Excluded: Unsafe research classifications

1,580 mapped genes | Alphabetical catalog with a PDF bookmark for every gene

Every entry carries its current classification, cancer occurrence, normal-expression evidence and curated biology. The complete tables retain unabridged annotations, all tumor cohorts, every normal context and source links.

Cancer RNA summaries rank cohorts by the fraction >=5 pTPM (at least 20 measured samples). Cancer IHC summaries rank Medium/High patient counts (at least five scored patients). These are different cohorts and native measures, not assay sensitivity estimates. No values are imputed for missing genes.

Biology excerpts come from reviewed UniProt entries (CC BY 4.0). They preserve qualifications but are not an independent review of every underlying paper. Clinical outcomes are reviewed only for the explicitly marked targets; a trial link alone establishes neither efficacy nor safety.

Read alongside the overview for the threshold grid, priority reconsideration list, known therapeutic risks, assay limitations and unmapped BAGE/GAGE2B records.

Open the searchable atlas and full tables

AANAT | Unsafe / RNA

ENSG00000129673

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.4 nTPM (limit 2); retina: 10.6 nTPM (limit 2); choroid plexus: 2.4 nTPM (baseline 2); retina: 10.6 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 4/176 (2.3%); Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 5/283 (1.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Controls the night/day rhythm of melatonin production in the pineal gland. Catalyzes the N-acetylation of serotonin into N-acetylserotonin, the penultimate step in the synthesis of melatonin

Curated tissue note. Highly expressed in pineal gland and at lower levels in the retina. Weak expression in several brain regions and in the pituitary gland

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q16613 | HPA portal (live; not the pinned values)

ABCA13 | Unsafe / RNA

ENSG00000179869

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 44.1 nTPM (limit 2); bone marrow: 44.1 nTPM (baseline 2)

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 39/68 (57.4%); Lung Squamous Cell Carcinoma (TCGA): 273/489 (55.8%); Glioblastoma Multiforme (validation): 21/58 (36.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May mediate the cholesterol and gangliosides transport from the plasma membrane to intracellular vesicles in an ATP hydrolysis dependent manner, thus playing a role in their internalization by endocytic retrograde transport and may also participate in the endocytosis of synaptic vesicle in cortical neurons

Curated tissue note. Significantly expressed in the bone marrow, trachea, testis, thyroid and lung as well as in skin fibroblasts

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86UQ4 | HPA portal (live; not the pinned values)

ABCA4 | Unsafe / Protein

ENSG00000198691

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / collecting ducts: High (Enhanced); kidney / distal tubules: High (Enhanced); kidney / proximal tubules (cell body): High (Enhanced); retina / photoreceptor cells: High (Enhanced)

Cancer RNA. Lung Adenocarcinoma (TCGA): 147/497 (29.6%); Lung Adenocarcinoma (validation): 29/105 (27.6%); Lung Squamous Cell Carcinoma (TCGA): 89/489 (18.2%)

Cancer IHC. renal cancer: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. Flippase that catalyzes in an ATP-dependent manner the transport of retinal-phosphatidylethanolamine conjugates like 11-cis and all-trans isomers of N-retinylidene-phosphatidylethanolamine (N-Ret-PE) from the lumen to the cytoplasmic leaflet of photoreceptor outer segment disk membranes, where 11-cis-retinylidene-phosphatidylethanolamine is then isomerized to its all-trans isomer and reduced by RDH8 to produce all-trans-retinol. This ... [full text in table]

Curated tissue note. Retinal-specific. Seems to be exclusively found in the rims of rod photoreceptor cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P78363 | HPA portal (live; not the pinned values)

ABHD1 | Unsafe / Protein

ENSG00000143994

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 25/50 (50.0%); Liver Hepatocellular Carcinoma (validation): 52/231 (22.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 11/282 (3.9%)

Cancer IHC. ovarian cancer: 11/11 (100.0%); endometrial cancer: 10/10 (100.0%)

Biology. Ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96SE0 | HPA portal (live; not the pinned values)

ABRAXAS1 | Unsafe / Protein

ENSG00000163322

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); colon / glandular cells: Medium (Uncertain); rectum / glandular cells: Medium (Uncertain); small intestine / glandular cells: Low (Uncertain); stomach 2 / glandular cells: Low (Uncertain)

Cancer RNA. Colon Adenocarcinoma (validation): 485/486 (99.8%); Rectum Adenocarcinoma (validation): 206/207 (99.5%); Glioblastoma Multiforme (validation): 57/58 (98.3%)

Cancer IHC. lymphoma: 1/11 (9.1%); testis cancer: 1/12 (8.3%)

Biology. Involved in DNA damage response and double-strand break (DSB) repair. Component of the BRCA1-A complex, acting as a central scaffold protein that assembles the various components of the complex and mediates the recruitment of BRCA1. The BRCA1-A complex specifically recognizes 'Lys-63'-linked ubiquitinated histones H2A and H2AX at DNA lesion sites, leading to target the BRCA1-BARD1 heterodimer to sites of DNA damage at DSBs. This ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6UWZ7 | HPA portal (live; not the pinned values)

ACAD11 | Unsafe / Protein

ENSG00000240303

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Enhanced); cerebellum / granular cells - cytoplasm/membrane: Medium (Enhanced); cerebellum / processes in granular layer: High (Enhanced); cerebellum / processes in white matter: High (Enhanced); cerebellum / Purkinje cells - cytoplasm/membrane: Low (Enhanced); ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 502/521 (96.4%)

Cancer IHC. renal cancer: 4/12 (33.3%); liver cancer: 3/12 (25.0%)

Biology. Participates in fatty acid beta-oxidation (FAO) and energy production in the central nervous system (CNS), and to a lesser extent in the liver (PubMed:21237683). Catalyzes the first step in FAO, which consists in the proR-proR stereospecific alpha,beta-dehydrogenation of fatty acyl-CoA thioesters using the electron transfer flavoprotein (ETF) as their physiologic electron acceptor, with maximal activity towards saturated docosanoyl-CoA ... [full text in table]

Curated tissue note. Widely expressed with highest levels in brain (white matter of cerebellum) followed by liver, heart and kidney

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q709F0 | HPA portal (live; not the pinned values)

ACE2 | Unsafe / Protein

ENSG00000130234

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); colon / enterocytes - Microvilli: High (Enhanced); duodenum / enterocytes: Low (Enhanced); duodenum / enterocytes - Microvilli: High (Enhanced); kidney / bowman's capsule: Medium (Enhanced); kidney / proximal tubules (cell body): ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 181/207 (87.4%); Kidney Renal Papillary Cell Carcinoma (TCGA): 244/282 (86.5%); Kidney Renal Clear Cell Carcinoma (validation): 82/100 (82.0%)

Cancer IHC. renal cancer: 9/11 (81.8%); colorectal cancer: 8/12 (66.7%)

Biology. Essential counter-regulatory carboxypeptidase of the renin-angiotensin hormone system that is a critical regulator of blood volume, systemic vascular resistance, and thus cardiovascular homeostasis (PubMed:27217402). Converts angiotensin I to angiotensin 1-9, a nine-amino acid peptide with anti-hypertrophic effects in cardiomyocytes, and angiotensin II to angiotensin 1-7, which then acts as a beneficial vasodilator and ... [full text in table]

Curated tissue note. Expressed in endothelial cells from small and large arteries, and in arterial smooth muscle cells (at protein level) (PubMed:15141377). Expressed in enterocytes of the small intestine, Leydig cells and Sertoli ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BYF1 | HPA portal (live; not the pinned values)

ACOXL | Unsafe / RNA

ENSG00000153093

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. lung: 8.9 nTPM (limit 2); lung: 8.9 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 84/133 (63.2%); Prostate Adenocarcinoma (TCGA): 181/480 (37.7%); Lung Adenocarcinoma (validation): 37/105 (35.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NUZ1 | HPA portal (live; not the pinned values)

ACRBP | Unsafe / High RNA

ENSG00000111644

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 5316 nTPM; pbmc/platelets; hpa_single_cell_tissue; vital; 56.54 TPM; blood/non-classical monocyte; hpa_immune_cell_samples; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 39/50 (78.0%); Thyroid Carcinoma (TCGA): 278/495 (56.2%); Kidney Renal Clear Cell Carcinoma (validation): 48/100 (48.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Acrosomal protein that maintains proacrosin (pro-ACR) as an enzymatically inactive zymogen in the acrosome. Involved also in the acrosome formation

Curated tissue note. Expression restricted to testis in normal tissue. Expressed in a wide spectrum of cancers, including bladder, breast, liver, lung and colon cancers

Reconsideration. Resolve normal-cell RNA identity and reliable local protein evidence before changing the general exclusion.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEB7 | HPA portal (live; not the pinned values)

ACRV1 | Unsafe / RNA

ENSG00000134940

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. basal ganglia: 2.4 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (validation): 6/58 (10.3%); Glioblastoma Multiforme (TCGA): 9/141 (6.4%); Skin Cuteneous Melanoma (TCGA): 5/99 (5.1%)

Cancer IHC. melanoma: 1/10 (10.0%); breast cancer: 0/12 (0.0%)

Biology. Testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P26436 | HPA portal (live; not the pinned values)

ACSM1 | Unsafe / RNA

ENSG00000166743

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 3.5 nTPM (limit 2); liver: 9.8 nTPM (limit 2); bone marrow: 3.5 nTPM (baseline 2); liver: 9.8 nTPM (baseline 2)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 422/480 (87.9%); Liver Hepatocellular Carcinoma (validation): 163/231 (70.6%); Liver Hepatocellular Carcinoma (TCGA): 229/362 (63.3%)

Cancer IHC. prostate cancer: 3/10 (30.0%); glioma: 2/9 (22.2%)

Biology. Catalyzes the activation of fatty acids by CoA to produce an acyl-CoA, the first step in fatty acid metabolism (PubMed:10434065). Capable of activating medium-chain fatty acids (e.g. butyric (C4) to decanoic (C10) acids), and certain carboxylate-containing xenobiotics, e.g. benzoate (PubMed:10434065). Also catalyzes the activation of lipoate to lipoyl-nucleoside monophosphate (By similarity). Activates lipoate with GTP at a 1000-fold ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q08AH1 | HPA portal (live; not the pinned values)

ACTRT2 | Unsafe / Protein

ENSG00000169717

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. small intestine / glandular cells: Low (Enhanced); stomach 1 / glandular cells: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Stomach Adenocarcinoma (TCGA): 1/346 (0.3%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/7 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TDY3 | HPA portal (live; not the pinned values)

ACTRT3 | Unsafe / RNA

ENSG00000184378

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. choroid plexus: 2.3 nTPM (limit 2)

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 51/283 (18.0%); Lung Squamous Cell Carcinoma (validation): 12/68 (17.6%); Uterine Corpus Endometrial Carcinoma (TCGA): 24/176 (13.6%)

Cancer IHC. skin cancer: 3/10 (30.0%); liver cancer: 2/10 (20.0%)

Biology. Ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BYD9 | HPA portal (live; not the pinned values)

ACVR2B | Unsafe / Protein

ENSG00000114739

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: Low (Approved); colon / peripheral nerve/ganglion: Low (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 44/50 (88.0%); Testicular Germ Cell Tumor (TCGA): 103/133 (77.4%)

Cancer IHC. testis cancer: 5/12 (41.7%); breast cancer: 4/11 (36.4%)

Biology. Transmembrane serine/threonine kinase activin type-2 receptor forming an activin receptor complex with activin type-1 serine/threonine kinase receptors (ACVR1, ACVR1B or ACVR1c). Transduces the activin signal from the cell surface to the cytoplasm and is thus regulating many physiological and pathological processes including neuronal differentiation and neuronal survival, hair follicle development and cycling, FSH production by the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13705 | HPA portal (live; not the pinned values)

ADAD2 | Unsafe / RNA

ENSG00000140955

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. basal ganglia: 2.1 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%); Glioblastoma Multiforme (validation): 1/58 (1.7%); Rectum Adenocarcinoma (TCGA): 1/88 (1.1%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Required for male fertility and normal male germ cell differentiation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCV1 | HPA portal (live; not the pinned values)

ADAM12 | Unsafe / High RNA

ENSG00000148848

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 179.9 nTPM; brain/inhibitory neurons; hpa_single_cell_tissue; vital; 26.76 TPM; blood/TH17; dice_th17; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 49/50 (98.0%); Lung Squamous Cell Carcinoma (validation): 65/68 (95.6%); Glioblastoma Multiforme (validation): 51/58 (87.9%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Involved in skeletal muscle regeneration, specifically at the onset of cell fusion. Also involved in macrophage-derived giant cells (MGC) and osteoclast formation from mononuclear precursors (By similarity)

Curated tissue note. Isoform 1 is expressed in placenta and skeletal, cardiac, and smooth muscle. Isoform 2 seems to be expressed only in placenta or in embryo and fetus. Both forms were expressed in some tumor cells lines. Not ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43184 | HPA portal (live; not the pinned values)

ADAM20 | Unsafe / High RNA

ENSG00000134007

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 53.4 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 1.708 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Glioblastoma Multiforme (validation): 22/58 (37.9%); Pancreatic Adenocarcinoma (validation): 2/80 (2.5%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. breast cancer: 0/9 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. May be involved in sperm maturation and/or fertilization

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43506 | HPA portal (live; not the pinned values)

ADAM32 | Unsafe / High RNA

ENSG00000197140

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 50.1 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 4.657 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Glioblastoma Multiforme (validation): 42/58 (72.4%); Pancreatic Adenocarcinoma (validation): 29/80 (36.2%); Breast Invasive Carcinoma (validation): 16/50 (32.0%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May play a role in sperm development and fertilization This is a non-catalytic metalloprotease-like protein

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TC27 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ADAMTS17 | Unsafe / RNA

ENSG00000140470

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.1 nTPM (limit 2); liver: 7.4 nTPM (limit 2); pancreas: 4.8 nTPM (limit 2); skeletal muscle: 5.4 nTPM (limit 2); thymus: 10.4 nTPM (limit 10); choroid plexus: 2.1 nTPM (baseline 2); liver: 7.4 nTPM (baseline 2); pancreas: 4.8 nTPM (baseline 2); skeletal muscle: 5.4 nTPM (baseline ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 18/50 (36.0%); Liver Hepatocellular Carcinoma (validation): 43/231 (18.6%); Glioblastoma Multiforme (validation): 7/58 (12.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Extracellular metalloprotease that regulates skeletal development by modulating BMP-SMAD signaling and extracellular matrix microfibril organization, which controls chondrocyte differentiation and bone growth

Curated tissue note. Isoform 1 and isoform 2 are expressed at high levels in the lung, brain, whole eye and retina. Isoform 1 shows a weaker expression in the heart, kidney and skeletal muscle. Isoform 2 shows a weaker expression ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TE56 | HPA portal (live; not the pinned values)

ADAMTS20 | Unsafe / High RNA

ENSG00000173157

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 100.2 nTPM; brain/inhibitory neurons; hpa_single_cell_tissue; vital; 0.3 TPM; blood/T-reg; hpa_immune_monaco; vital

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 6/68 (8.8%); Glioblastoma Multiforme (validation): 4/58 (6.9%); Testicular Germ Cell Tumor (TCGA): 5/133 (3.8%)

Cancer IHC. testis cancer: 11/12 (91.7%); pancreatic cancer: 10/12 (83.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

HPA portal (live; not the pinned values)

ADAMTS6 | Unsafe / High RNA

ENSG00000049192

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 243.5 nTPM; brain/inhibitory neurons; hpa_single_cell_tissue; vital; 10.45 TPM; blood/CD8_NAIVE; dice_cd8_naive; vital

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Pancreatic Adenocarcinoma (validation): 51/80 (63.7%); Breast Invasive Carcinoma (validation): 22/50 (44.0%)

Cancer IHC. pancreatic cancer: 1/8 (12.5%); breast cancer: 0/11 (0.0%)

Biology. Expressed at low levels in placenta and barely detectable in a number of other tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UKP5 | HPA portal (live; not the pinned values)

ADCY10 | Unsafe / Protein

ENSG00000143199

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebral cortex / glial cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); cerebral cortex / neuropil: Low (Approved); colon / ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (TCGA): 85/362 (23.5%); Liver Hepatocellular Carcinoma (validation): 31/231 (13.4%); Breast Invasive Carcinoma (validation): 2/50 (4.0%)

Cancer IHC. endometrial cancer: 2/12 (16.7%); glioma: 2/12 (16.7%)

Biology. Catalyzes the formation of the signaling molecule cAMP (PubMed:12609998, PubMed:15659711, PubMed:24567411, PubMed:24616449, PubMed:25040695). May function as sensor that mediates responses to changes in cellular bicarbonate and CO(2) levels (PubMed:15659711, PubMed:17591988). Has a critical role in mammalian spermatogenesis by producing the cAMP which regulates cAMP-responsive nuclear factors indispensable for sperm maturation in the ... [full text in table]

Curated tissue note. Detected in airway epithelial cells and testis (at protein level) (PubMed:17591988). Weakly expressed in multiple tissues. Expressed in brain, heart, kidney, liver, lung, pancreas, peripheral blood leukocytes, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96PN6 | HPA portal (live; not the pinned values)

ADGB | Unsafe / Protein

ENSG00000118492

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cell body): Low (Enhanced); nasopharynx / ciliated cells (cilia axoneme): High (Enhanced); ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 11/176 (6.2%); Glioblastoma Multiforme (validation): 2/58 (3.4%); Ovary Serous Cystadenocarcinoma (validation): 1/81 (1.2%)

Cancer IHC. renal cancer: 3/11 (27.3%); testis cancer: 2/9 (22.2%)

Biology. Probable chimeric globin with a bis-histidyl six-coordinate heme-iron atom through which it could bind dioxygen, carbon monoxide and nitric oxide (PubMed:22115833). Required for sperm flagellum formation and maturation of elongating spermatids, thus playing an essential role in male fertility (PubMed:36995441)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N7X0 | HPA portal (live; not the pinned values)

ADORA2A | Unsafe / Protein

ENSG00000128271

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); caudate / neuronal projections: High (Enhanced); caudate / synapses: High (Enhanced); cerebellum / molecular layer cells - cytoplasm/membrane: Medium (Enhanced); cerebellum / processes in white ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 121/133 (91.0%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Kidney Renal Clear Cell Carcinoma (validation): 88/100 (88.0%)

Cancer IHC. ovarian cancer: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. Receptor for adenosine (By similarity). The activity of this receptor is mediated by G proteins which activate adenylyl cyclase (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P29274 | HPA portal (live; not the pinned values)

AFF4 | Unsafe / Protein

ENSG00000072364

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / ciliated cells (cell body): Low (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: Low (Approved); cerebellum ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. glioma: 10/12 (83.3%); skin cancer: 6/11 (54.5%)

Biology. Key component of the super elongation complex (SEC), a complex required to increase the catalytic rate of RNA polymerase II transcription by suppressing transient pausing by the polymerase at multiple sites along the DNA. In the SEC complex, AFF4 acts as a central scaffold that recruits other factors through direct interactions with ELL proteins (ELL, ELL2 or ELL3) and the P-TEFb complex. In case of infection by HIV-1 virus, the SEC ... [full text in table]

Curated tissue note. Ubiquitously expressed. Strongly expressed in heart, placenta, skeletal muscle, pancreas and to a lower extent in brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UHB7 | HPA portal (live; not the pinned values)

AFG1L | Unsafe / Protein

ENSG00000135537

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / ciliated cells (cell body): High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Colon Adenocarcinoma (validation): 333/486 (68.5%); Rectum Adenocarcinoma (validation): 110/207 (53.1%)

Cancer IHC. breast cancer: 9/12 (75.0%); ovarian cancer: 7/12 (58.3%)

Biology. Putative mitochondrial ATPase. Plays a role in mitochondrial morphology and mitochondrial protein metabolism. Promotes degradation of excess nuclear-encoded complex IV subunits (COX4I1, COX5A and COX6A1) and normal activity of complexes III and IV of the respiratory chain (PubMed:26759378, PubMed:27323408). Mediates mitochondrial translocation of TP53 and its transcription-independent apoptosis in response to genotoxic stress ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WV93 | HPA portal (live; not the pinned values)

AFG2A | Unsafe / Protein

ENSG00000145375

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 77/80 (96.2%); Colon Adenocarcinoma (validation): 415/486 (85.4%)

Cancer IHC. glioma: 12/12 (100.0%); testis cancer: 11/11 (100.0%)

Biology. ATP-dependent chaperone part of the 55LCC heterohexameric ATPase complex which is chromatin-associated and promotes replisome proteostasis to maintain replication fork progression and genome stability. Required for replication fork progression, sister chromatid cohesion, and chromosome stability. The ATPase activity is specifically enhanced by replication fork DNA and is coupled to cysteine protease-dependent cleavage of replisome ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NB90 | HPA portal (live; not the pinned values)

AGBL2 | Unsafe / Protein

ENSG00000165923

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); caudate / glial cells: High (Uncertain); caudate / neuronal cells: High (Uncertain); cerebellum / cells in granular layer: High (Uncertain); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 28/50 (56.0%); Ovary Serous Cystadenocarcinoma (validation): 12/81 (14.8%); Liver Hepatocellular Carcinoma (validation): 31/231 (13.4%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Metallocarboxypeptidase that mediates deglutamylation of tubulin and non-tubulin target proteins. Catalyzes the removal of polyglutamate side chains present on the gamma-carboxyl group of glutamate residues within the C-terminal tail of tubulin protein. Specifically cleaves tubulin long-side-chains, while it is not able to remove the branching point glutamate. Also catalyzes the removal of polyglutamate residues from the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5U5Z8 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

AGBL5 | Unsafe / Protein

ENSG00000084693

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / cells in ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Colon Adenocarcinoma (validation): 486/486 (100.0%); Ovary Serous Cystadenocarcinoma (TCGA): 349/349 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); melanoma: 11/11 (100.0%)

Biology. Metallocarboxypeptidase that mediates deglutamylation of tubulin and non-tubulin target proteins. Catalyzes the removal of polyglutamate side chains present on the gamma-carboxyl group of glutamate residues within the C-terminal tail of alpha- and beta-tubulin. Cleaves alpha- and gamma-linked polyglutamate tubulin side-chain, as well as the branching point glutamate. Also catalyzes the removal of alpha-linked glutamate residues from ... [full text in table]

Curated tissue note. Expressed in brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NDL9 | HPA portal (live; not the pinned values)

AHCTF1 | Unsafe / Protein

ENSG00000153207

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Required for the assembly of a functional nuclear pore complex (NPC) on the surface of chromosomes as nuclei form at the end of mitosis. May initiate NPC assembly by binding to chromatin and recruiting the Nup107-160 subcomplex of the NPC. Also required for the localization of the Nup107-160 subcomplex of the NPC to the kinetochore during mitosis and for the completion of cytokinesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WYP5 | HPA portal (live; not the pinned values)

AHRR | Unsafe / RNA

ENSG00000063438

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. cerebral cortex: 2.1 nTPM (limit 2); lung: 2.1 nTPM (limit 2); cerebral cortex: 2.1 nTPM (baseline 2); lung: 2.1 nTPM (baseline 2)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 29/99 (29.3%); Breast Invasive Carcinoma (validation): 12/50 (24.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 23/283 (8.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Mediates dioxin toxicity and is involved in regulation of cell growth and differentiation. Represses the transcription activity of AHR by competing with AHR for heterodimer formation with ARNT and subsequently binding to the xenobiotic response element (XRE) sequence present in the promoter regulatory region of a variety of genes. Represses CYP1A1 by binding the XRE sequence and recruiting ANKRA2, HDAC4 and/or HDAC5. Autoregulates its ... [full text in table]

Curated tissue note. Highly expressed in testis, lung, ovary, spleen and pancreas. Highly expressed in mononuclear cells (MNCs) from umbilical cord blood. Down-regulated in malignant tissue from different anatomical origins, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A9YTQ3 | HPA portal (live; not the pinned values)

AIPL1 | Unsafe / Protein

ENSG00000129221

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: High (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 27/133 (20.3%); Pancreatic Adenocarcinoma (validation): 3/80 (3.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 4/176 (2.3%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May be important in protein trafficking and/or protein folding and stabilization

Curated tissue note. Highly expressed in retina. Specifically localized to the developing photoreceptor layer and within the photoreceptors of the adult retina

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NZN9 | HPA portal (live; not the pinned values)

AK7 | Unsafe / Protein

ENSG00000140057

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); duodenum / paneth cells: High (Enhanced); kidney / cells in glomeruli: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); lung / macrophages: Low (Enhanced); ... [full text in table]

Cancer RNA. Kidney Renal Papillary Cell Carcinoma (TCGA): 201/282 (71.3%); Uterine Corpus Endometrial Carcinoma (TCGA): 82/176 (46.6%); Breast Invasive Carcinoma (validation): 22/50 (44.0%)

Cancer IHC. prostate cancer: 11/11 (100.0%); breast cancer: 10/12 (83.3%)

Biology. Nucleoside monophosphate (NMP) kinase that catalyzes the reversible transfer of the terminal phosphate group between nucleoside triphosphates and monophosphates. Has highest activity toward AMP, and weaker activity toward dAMP, CMP and dCMP. Also displays broad nucleoside diphosphate kinase activity. Involved in maintaining ciliary structure and function

Curated tissue note. Expressed in sperm and airway epithelial cells (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96M32 | HPA portal (live; not the pinned values)

AK8 | Unsafe / Protein

ENSG00000165695

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); kidney / distal tubules: Low (Enhanced); nasopharynx / ciliated cells (cilia axoneme): High (Enhanced); nasopharynx / ciliated cells (tip of cilia): High (Enhanced)

Cancer RNA. Kidney Renal Papillary Cell Carcinoma (TCGA): 187/282 (66.3%); Thyroid Carcinoma (TCGA): 309/495 (62.4%); Glioblastoma Multiforme (validation): 29/58 (50.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Nucleoside monophosphate (NMP) kinase that catalyzes the reversible transfer of the terminal phosphate group between nucleoside triphosphates and monophosphates. Has highest activity toward AMP, and weaker activity toward dAMP, CMP and dCMP. Also displays broad nucleoside diphosphate kinase activity

Curated tissue note. Expressed in respiratory cells (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96MA6 | HPA portal (live; not the pinned values)

AK9 | Unsafe / Protein

ENSG00000155085

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Enhanced); adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: High (Enhanced); bronchus / respiratory epithelial cells: High (Enhanced); caudate / glial cells: High (Enhanced); cerebellum / cells in granular layer: High ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 470/486 (96.7%); Rectum Adenocarcinoma (validation): 200/207 (96.6%); Pancreatic Adenocarcinoma (validation): 63/80 (78.8%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); lung cancer: 12/12 (100.0%)

Biology. Broad-specificity nucleoside phosphate kinase involved in cellular nucleotide homeostasis by catalyzing nucleoside-phosphate interconversions. Similar to other adenylate kinases, preferentially catalyzes the phosphorylation of the nucleoside monophosphate AMP with ATP as phosphate donor to produce ADP. In vitro, can also catalyze the phosphorylation of CMP, dAMP and dCMP and use GTP as an alternate phosphate donor. Moreover, exhibits a ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5TCS8 | HPA portal (live; not the pinned values)

AKAP14 | Unsafe / Protein

ENSG00000186471

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Low (Enhanced); bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); nasopharynx / ciliated cells (cilia axoneme): Medium (Enhanced); nasopharynx / ciliated cells (tip of cilia): Medium ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 34/176 (19.3%); Breast Invasive Carcinoma (validation): 7/50 (14.0%); Lung Adenocarcinoma (TCGA): 48/497 (9.7%)

Cancer IHC. prostate cancer: 2/12 (16.7%); melanoma: 1/12 (8.3%)

Biology. Binds to type II regulatory subunits of protein kinase A and anchors/targets them

Curated tissue note. Present in cilia (at protein level). Expressed in tissues containing axoneme-based organelles (cilia and/or flagella): trachea and testis. Highly expressed in airway cilia

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86UN6 | HPA portal (live; not the pinned values)

AKAP3 | Unsafe / RNA

ENSG00000111254

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. spinal cord: 2.2 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 63/133 (47.4%); Glioblastoma Multiforme (validation): 20/58 (34.5%); Breast Invasive Carcinoma (validation): 11/50 (22.0%)

Cancer IHC. colorectal cancer: 1/10 (10.0%); breast cancer: 1/11 (9.1%)

Biology. Structural component of sperm fibrous sheath (By similarity). Required for the formation of the subcellular structure of the sperm flagellum, sperm motility and male fertility (PubMed:35228300)

Curated tissue note. Testis specific; only expressed in spermatids

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75969 | HPA portal (live; not the pinned values)

AKIRIN2 | Unsafe / RNA

ENSG00000135334

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 31.8 nTPM (limit 10); adrenal gland: 35.9 nTPM (limit 2); amygdala: 33.9 nTPM (limit 2); basal ganglia: 33.1 nTPM (limit 2); bone marrow: 69.6 nTPM (limit 2); cerebellum: 27.4 nTPM (limit 2); cerebral cortex: 35.8 nTPM (limit 2); choroid plexus: 24.4 nTPM (limit 2); colon: 19.2 nTPM ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Molecular adapter that acts as a bridge between a variety of multiprotein complexes, and which is involved in embryonic development, immunity, myogenesis and brain development (PubMed:34711951). Plays a key role in nuclear protein degradation by promoting import of proteasomes into the nucleus: directly binds to fully assembled 20S proteasomes at one end and to nuclear import receptor IPO9 at the other end, bridging them together and ... [full text in table]

Curated tissue note. Widely expressed with the highest expression in peripheral blood leukocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q53H80 | HPA portal (live; not the pinned values)

AKR1E2 | Unsafe / Protein

ENSG00000165568

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); cerebral cortex / glial cells: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); colon / glandular cells: Low (Enhanced); duodenum / glandular cells: Low (Enhanced); kidney / ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 338/486 (69.5%); Breast Invasive Carcinoma (validation): 33/50 (66.0%); Rectum Adenocarcinoma (validation): 127/207 (61.4%)

Cancer IHC. renal cancer: 3/11 (27.3%); breast cancer: 2/11 (18.2%)

Biology. Catalyzes the NADPH-dependent reduction of 1,5-anhydro-D-fructose (AF) to 1,5-anhydro-D-glucitol (By similarity). Has low NADPH-dependent reductase activity towards 9,10-phenanthrenequinone (in vitro) (PubMed:12604216, PubMed:15118078)

Curated tissue note. Specifically expressed in testis (PubMed:12604216, PubMed:15118078). Expressed in testicular germ cells and testis interstitial cells (PubMed:15118078)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96JD6 | HPA portal (live; not the pinned values)

ALG10 | Unsafe / Protein

ENSG00000139133

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: High ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 188/207 (90.8%); Colon Adenocarcinoma (validation): 439/486 (90.3%)

Cancer IHC. breast cancer: 10/11 (90.9%); cervical cancer: 10/12 (83.3%)

Biology. Dol-P-Glc:Glc(2)Man(9)GlcNAc(2)-PP-Dol alpha-1,2-glucosyltransferase that operates in the biosynthetic pathway of dolichol-linked oligosaccharides, the glycan precursors employed in protein asparagine (N)-glycosylation. The assembly of dolichol-linked oligosaccharides begins on the cytosolic side of the endoplasmic reticulum membrane and finishes in its lumen. The sequential addition of sugars to dolichol pyrophosphate produces ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5BKT4 | HPA portal (live; not the pinned values)

ALG10B | Unsafe / Protein

ENSG00000175548

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: High ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 42/50 (84.0%); Pancreatic Adenocarcinoma (validation): 66/80 (82.5%)

Cancer IHC. breast cancer: 10/11 (90.9%); cervical cancer: 10/12 (83.3%)

Biology. Dol-P-Glc:Glc(2)Man(9)GlcNAc(2)-PP-Dol alpha-1,2-glucosyltransferase that operates in the biosynthetic pathway of dolichol-linked oligosaccharides, the glycan precursors employed in protein asparagine (N)-glycosylation. The assembly of dolichol-linked oligosaccharides begins on the cytosolic side of the endoplasmic reticulum membrane and finishes in its lumen. The sequential addition of sugars to dolichol pyrophosphate produces ... [full text in table]

Curated tissue note. Highly expressed in heart, placenta, liver, kidney and pancreas. Weakly expressed in lung, skeletal muscle and brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5I7T1 | HPA portal (live; not the pinned values)

ALK | Unsafe / Protein

ENSG00000171094

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: High ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 32/58 (55.2%); Skin Cuteneous Melanoma (TCGA): 12/99 (12.1%); Glioblastoma Multiforme (TCGA): 17/141 (12.1%)

Cancer IHC. prostate cancer: 7/12 (58.3%); skin cancer: 5/10 (50.0%)

Biology. Neuronal receptor tyrosine kinase that is essentially and transiently expressed in specific regions of the central and peripheral nervous systems and plays an important role in the genesis and differentiation of the nervous system (PubMed:11121404, PubMed:11387242, PubMed:16317043, PubMed:17274988, PubMed:30061385, PubMed:34646012, PubMed:34819673). Also acts as a key thinness protein involved in the resistance to weight gain: in ... [full text in table]

Curated tissue note. Expressed in brain and CNS. Also expressed in the small intestine and testis, but not in normal lymphoid cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UM73 | HPA portal (live; not the pinned values)

ALPG | Unsafe / Protein

ENSG00000163286

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / enterocytes - Microvilli: Medium (Approved); duodenum / enterocytes - Microvilli: High (Approved); rectum / enterocytes - Microvilli: High (Approved); skin 1 / fibrohistiocytic cells: Low (Approved); small intestine / enterocytes - Microvilli: High (Approved)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 118/133 (88.7%); Uterine Corpus Endometrial Carcinoma (TCGA): 63/176 (35.8%); Ovary Serous Cystadenocarcinoma (TCGA): 84/349 (24.1%)

Cancer IHC. testis cancer: 9/11 (81.8%); endometrial cancer: 5/12 (41.7%)

Biology. Alkaline phosphatase that can hydrolyze various phosphate compounds

Curated tissue note. Trace amounts in the testis and thymus, and in elevated amounts in germ cell tumors

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P10696 | HPA portal (live; not the pinned values)

ALPP | Unsafe / Protein

ENSG00000163283

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / enterocytes - Microvilli: Medium (Enhanced); duodenum / enterocytes - Microvilli: High (Enhanced); rectum / enterocytes - Microvilli: High (Enhanced); skin 1 / fibrohistiocytic cells: Low (Enhanced); small intestine / enterocytes - Microvilli: High (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 60/133 (45.1%); Uterine Corpus Endometrial Carcinoma (TCGA): 55/176 (31.2%); Ovary Serous Cystadenocarcinoma (validation): 20/81 (24.7%)

Cancer IHC. testis cancer: 9/11 (81.8%); endometrial cancer: 5/12 (41.7%)

Biology. Alkaline phosphatase that can hydrolyze various phosphate compounds

Curated tissue note. Detected in placenta (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P05187 | HPA portal (live; not the pinned values)

AMER1 | Unsafe / Protein

ENSG00000184675

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Rectum Adenocarcinoma (validation): 165/207 (79.7%); Colon Adenocarcinoma (validation): 372/486 (76.5%)

Cancer IHC. lymphoma: 12/12 (100.0%); cervical cancer: 11/11 (100.0%)

Biology. Regulator of the canonical Wnt signaling pathway. Acts by specifically binding phosphatidylinositol 4,5-bisphosphate (PtdIns(4,5)P2), translocating to the cell membrane and interacting with key regulators of the canonical Wnt signaling pathway, such as components of the beta-catenin destruction complex. Acts both as a positive and negative regulator of the Wnt signaling pathway, depending on the context: acts as a positive regulator by ... [full text in table]

Curated tissue note. Detected in fetal and adult kidney, brain and spleen

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JTC6 | HPA portal (live; not the pinned values)

AMIGO3 | Unsafe / RNA

ENSG00000176020

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pituitary gland: 3 nTPM (limit 2); skeletal muscle: 3.3 nTPM (limit 2); pituitary gland: 3 nTPM (baseline 2); skeletal muscle: 3.3 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 37/50 (74.0%); Liver Hepatocellular Carcinoma (validation): 82/231 (35.5%); Uterine Corpus Endometrial Carcinoma (TCGA): 37/176 (21.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May mediate heterophilic cell-cell interaction. May contribute to signal transduction through its intracellular domain (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86WK7 | HPA portal (live; not the pinned values)

ANG | Unsafe / RNA

ENSG00000214274

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 36.4 nTPM (limit 10); adrenal gland: 2.9 nTPM (limit 2); amygdala: 2.5 nTPM (limit 2); basal ganglia: 2.7 nTPM (limit 2); cerebral cortex: 2.2 nTPM (limit 2); choroid plexus: 23.3 nTPM (limit 2); colon: 31.5 nTPM (limit 6); duodenum: 22.5 nTPM (limit 2); esophagus: 9.5 nTPM (limit ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (TCGA): 362/362 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%); Rectum Adenocarcinoma (TCGA): 88/88 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Secreted ribonuclease that can either promote or restrict cell proliferation of target cells, depending on the context (PubMed:12051708, PubMed:1400510, PubMed:19332886, PubMed:20129916, PubMed:21855800, PubMed:23047679, PubMed:23843625, PubMed:2424496, PubMed:2459697, PubMed:2730651, PubMed:27518564, PubMed:28176817, PubMed:29100074, PubMed:29748193, PubMed:3122207, PubMed:32510170, PubMed:38718836, PubMed:8159680, PubMed:8570639, ... [full text in table]

Curated tissue note. Expressed predominantly in the liver (PubMed:2440105). Also detected in endothelial cells and spinal cord neurons (PubMed:17886298, PubMed:2440105)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P03950 | HPA portal (live; not the pinned values)

ANKAR | Unsafe / High RNA

ENSG00000151687

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 81.6 nTPM; unsited/Oligodendrocyte precursor cells; hpa_single_cell; unresolved; 30.74 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Pancreatic Adenocarcinoma (validation): 52/80 (65.0%); Breast Invasive Carcinoma (validation): 24/50 (48.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); liver cancer: 11/11 (100.0%)

Biology. Ubiquitously expressed with highest level in pancreas and lowest in skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z5J8 | HPA portal (live; not the pinned values)

ANKRD33 | Unsafe / Protein

ENSG00000167612

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: High (Enhanced)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 6/231 (2.6%); Liver Hepatocellular Carcinoma (TCGA): 6/362 (1.7%); Ovary Serous Cystadenocarcinoma (validation): 1/81 (1.2%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Acts as a transcriptional repressor for CRX-activated photoreceptor gene regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z3H0 | HPA portal (live; not the pinned values)

ANKRD36B | Unsafe / RNA

ENSG00000196912

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.2 nTPM (limit 2); basal ganglia: 2.5 nTPM (limit 2); bone marrow: 7.5 nTPM (limit 2); cerebellum: 8.9 nTPM (limit 2); cerebral cortex: 2.5 nTPM (limit 2); hippocampal formation: 2.6 nTPM (limit 2); hypothalamus: 2.1 nTPM (limit 2); lung: 2.1 nTPM (limit 2); midbrain: 2.2 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 79/80 (98.8%); Breast Invasive Carcinoma (validation): 45/50 (90.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N2N9 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ANKRD45 | Unsafe / Protein

ENSG00000183831

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cilia axoneme): Medium (Enhanced); nasopharynx / ciliated cells (tip of cilia): Medium (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 41/58 (70.7%); Ovary Serous Cystadenocarcinoma (validation): 20/81 (24.7%); Uterine Corpus Endometrial Carcinoma (TCGA): 34/176 (19.3%)

Cancer IHC. colorectal cancer: 1/10 (10.0%); breast cancer: 0/11 (0.0%)

Biology. May play a role during cell division

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5TZF3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ANKRD55 | Unsafe / Protein

ENSG00000164512

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebellum / Purkinje cells: Low (Uncertain); cerebral cortex / neuronal cells: Low ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 16/133 (12.0%); Breast Invasive Carcinoma (validation): 3/50 (6.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%)

Cancer IHC. prostate cancer: 9/11 (81.8%); lung cancer: 6/11 (54.5%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3KP44 | HPA portal (live; not the pinned values)

ANKRD62 | Unsafe / High RNA

ENSG00000181626

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 47.6 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 1.02 TPM; blood/NK; dice_nk; vital

Cancer RNA. Pancreatic Adenocarcinoma (validation): 2/80 (2.5%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. colorectal cancer: 3/10 (30.0%); endometrial cancer: 1/11 (9.1%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NC57 | HPA portal (live; not the pinned values)

ANKRD7 | Unsafe / High RNA

ENSG00000106013

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 34.1 nTPM; testis/macrophages; hpa_single_cell_tissue; vital; 2.05 TPM; blood/memory B-cell; hpa_immune_cell_samples; vital

Cancer RNA. Glioblastoma Multiforme (validation): 30/58 (51.7%); Testicular Germ Cell Tumor (TCGA): 15/133 (11.3%); Skin Cuteneous Melanoma (TCGA): 8/99 (8.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92527 | HPA portal (live; not the pinned values)

ANKS1B | Unsafe / Protein

ENSG00000185046

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Medium (Enhanced); hippocampus / neuronal cells: Low (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 40/50 (80.0%); Glioblastoma Multiforme (validation): 39/58 (67.2%); Glioblastoma Multiforme (TCGA): 92/141 (65.2%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Isoform 2 may participate in the regulation of nucleoplasmic coilin protein interactions in neuronal and transformed cells Isoform 3 can regulate global protein synthesis by altering nucleolar numbers Isoform 4 may play a role as a modulator of APP processing. Overexpression can down-regulate APP processing

Curated tissue note. Highly expressed in marrow from patients with pre-B ALL associated with the t(1;19) translocation. Strongly expressed in brain and testis. Expressed in fetal brain. Isoform 4 is highly expressed in brain (at ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z6G8 | HPA portal (live; not the pinned values)

ANO2 | Unsafe / Protein

ENSG00000047617

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): High (Enhanced); nasopharynx / ciliated cells (ciliary rootlets): Medium (Enhanced); retina / outer plexiform layer: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 88/133 (66.2%); Glioblastoma Multiforme (validation): 8/58 (13.8%); Ovary Serous Cystadenocarcinoma (TCGA): 36/349 (10.3%)

Cancer IHC. colorectal cancer: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. Calcium-activated chloride channel (CaCC) which may play a role in olfactory signal transduction. Odorant molecules bind to odor-sensing receptors (OSRs), leading to an increase in calcium entry that activates CaCC current which amplifies the depolarization of the OSR cells, ANO2 seems to be the underlying chloride channel involved in this process. May mediate light perception amplification in retina

Curated tissue note. Retina, especially in the photoreceptor synaptic terminals

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NQ90 | HPA portal (live; not the pinned values)

AP3B2 | Unsafe / Protein

ENSG00000103723

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / GLUC cells - cytoplasm/membrane: High (Enhanced); cerebellum / granular cells - cytoplasm/membrane: Medium (Enhanced); cerebellum / molecular layer - neuropil: Medium (Enhanced); cerebellum / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 47/58 (81.0%); Glioblastoma Multiforme (TCGA): 111/141 (78.7%); Breast Invasive Carcinoma (validation): 29/50 (58.0%)

Cancer IHC. colorectal cancer: 7/12 (58.3%); glioma: 1/11 (9.1%)

Biology. Subunit of non-clathrin- and clathrin-associated adaptor protein complex 3 (AP-3) that plays a role in protein sorting in the late-Golgi/trans-Golgi network (TGN) and/or endosomes. The AP complexes mediate both the recruitment of clathrin to membranes and the recognition of sorting signals within the cytosolic tails of transmembrane cargo molecules. AP-3 appears to be involved in the sorting of a subset of transmembrane proteins ... [full text in table]

Curated tissue note. Isoform 1 expression is specific to nervous system. Expressed in nerve terminal and cell body, and is associated with nerve-terminal vesicles. Expression seen in Purkinje cells, cortical neurons, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13367 | HPA portal (live; not the pinned values)

AP4S1 | Unsafe / Protein

ENSG00000100478

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: High (Approved); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 103/105 (98.1%); Rectum Adenocarcinoma (validation): 202/207 (97.6%); Glioblastoma Multiforme (validation): 56/58 (96.6%)

Cancer IHC. liver cancer: 12/12 (100.0%); testis cancer: 12/12 (100.0%)

Biology. Component of the adaptor protein complex 4 (AP-4). Adaptor protein complexes are vesicle coat components involved both in vesicle formation and cargo selection. They control the vesicular transport of proteins in different trafficking pathways (PubMed:10066790, PubMed:10436028). AP-4 forms a non clathrin-associated coat on vesicles departing the trans-Golgi network (TGN) and may be involved in the targeting of proteins from the ... [full text in table]

Curated tissue note. Widely expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y587 | HPA portal (live; not the pinned values)

APLF | Unsafe / High RNA

ENSG00000169621

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 32 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 9.709 TPM; blood/basophil; hpa_immune_cell_samples; vital

Cancer RNA. Glioblastoma Multiforme (validation): 53/58 (91.4%); Breast Invasive Carcinoma (validation): 36/50 (72.0%); Colon Adenocarcinoma (validation): 135/486 (27.8%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Histone chaperone involved in single-strand and double-strand DNA break repair (PubMed:17353262, PubMed:17396150, PubMed:21211721, PubMed:21211722, PubMed:29905837, PubMed:30104678). Recruited to sites of DNA damage through interaction with branched poly-ADP-ribose chains, a polymeric post-translational modification synthesized transiently at sites of chromosomal damage to accelerate DNA strand break repair reactions (PubMed:17353262, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IW19 | HPA portal (live; not the pinned values)

APOBEC3F | Unsafe / RNA

ENSG00000128394

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 6.1 nTPM (limit 2); bone marrow: 3.1 nTPM (limit 2); choroid plexus: 5.9 nTPM (limit 2); duodenum: 3.5 nTPM (limit 2); heart muscle: 5.3 nTPM (limit 2); kidney: 3.3 nTPM (limit 2); liver: 2.8 nTPM (limit 2); lung: 3.3 nTPM (limit 2); lymph node: 15.2 nTPM (limit 6); pancreas: 2.9 ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 477/486 (98.1%); Rectum Adenocarcinoma (validation): 203/207 (98.1%); Thyroid Carcinoma (TCGA): 440/495 (88.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. DNA deaminase (cytidine deaminase) which acts as an inhibitor of retrovirus replication and retrotransposon mobility via deaminase-dependent and -independent mechanisms (PubMed:15141007, PubMed:17121840, PubMed:20062055, PubMed:22915799, PubMed:34774569). Exhibits antiviral activity against viruse such as HIV-1 or HIV-2 (PubMed:15141007, PubMed:15152192, PubMed:17121840, PubMed:20219927, PubMed:21835787, PubMed:22807680, ... [full text in table]

Curated tissue note. Widely expressed. Highly expressed in ovary

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IUX4 | HPA portal (live; not the pinned values)

APOBEC4 | Unsafe / High RNA

ENSG00000173627

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 42.7 nTPM; lung/ciliated cells; hpa_single_cell_tissue; vital; 0.5108 TPM; blood/CD4_NAIVE; dice_cd4_naive; vital

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 16/176 (9.1%); Ovary Serous Cystadenocarcinoma (validation): 3/81 (3.7%); Lung Adenocarcinoma (TCGA): 10/497 (2.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Putative deaminase whose physiological substrate is not yet known

Curated tissue note. Predominantly expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WW27 | HPA portal (live; not the pinned values)

AQP5 | Unsafe / Protein

ENSG00000161798

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Enhanced); duodenum / glandular cells: Low (Enhanced); nasopharynx / respiratory epithelial cells: Medium (Enhanced); pancreas / exocrine glandular cells: Low (Enhanced); salivary gland / glandular cells: High (Enhanced); stomach 1 / glandular cells: ... [full text in table]

Cancer RNA. Ovary Serous Cystadenocarcinoma (validation): 75/81 (92.6%); Ovary Serous Cystadenocarcinoma (TCGA): 318/349 (91.1%); Pancreatic Adenocarcinoma (TCGA): 150/176 (85.2%)

Cancer IHC. endometrial cancer: 10/12 (83.3%); pancreatic cancer: 5/8 (62.5%)

Biology. Aquaporins form homotetrameric transmembrane channels, with each monomer independently mediating water transport across the plasma membrane along its osmotic gradient (PubMed:18768791, PubMed:8621489). Plays an important role in fluid secretion in salivary glands (By similarity). Required for TRPV4 activation by hypotonicity. Together with TRPV4, controls regulatory volume decrease in salivary epithelial cells (PubMed:16571723). Seems ... [full text in table]

Curated tissue note. Detected in skin eccrine sweat glands, at the apical cell membrane and at intercellular canaliculi (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P55064 | HPA portal (live; not the pinned values)

ARFGEF2 | Unsafe / Protein

ENSG00000124198

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Supported); adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: Medium (Supported); bronchus / respiratory epithelial cells: Medium (Supported); caudate / glial cells: Low (Supported); caudate / neuronal cells: Low (Supported); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 50/50 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); pancreatic cancer: 12/12 (100.0%)

Biology. Promotes guanine-nucleotide exchange on ARF1 and ARF3 and to a lower extent on ARF5 and ARF6. Promotes the activation of ARF1/ARF5/ARF6 through replacement of GDP with GTP. Involved in the regulation of Golgi vesicular transport. Required for the integrity of the endosomal compartment. Involved in trafficking from the trans-Golgi network (TGN) to endosomes and is required for membrane association of the AP-1 complex and GGA1. Seems to ... [full text in table]

Curated tissue note. Expressed in placenta, lung, heart, brain, kidney and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y6D5 | HPA portal (live; not the pinned values)

ARHGAP11A | Unsafe / Protein

ENSG00000198826

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in molecular layer: Medium (Enhanced); cerebellum / Purkinje cells: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 205/207 (99.0%); Glioblastoma Multiforme (validation): 57/58 (98.3%)

Cancer IHC. testis cancer: 11/11 (100.0%); melanoma: 10/11 (90.9%)

Biology. GTPase activator for the Rho-type GTPases by converting them to an inactive GDP-bound state

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6P4F7 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ARHGAP11B | Unsafe / RNA

ENSG00000285077

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 5 nTPM (limit 2); bone marrow: 5 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 36/50 (72.0%); Rectum Adenocarcinoma (validation): 5/207 (2.4%); Colon Adenocarcinoma (validation): 11/486 (2.3%)

Cancer IHC. liver cancer: 2/11 (18.2%); breast cancer: 0/11 (0.0%)

Biology. Hominin-specific protein that promotes development and evolutionary expansion of the brain neocortex (PubMed:25721503, PubMed:27957544, PubMed:30484771, PubMed:32554627, PubMed:33938018). Able to promote amplification of basal progenitors in the subventricular zone, producing more neurons during fetal corticogenesis, thereby playing a key role in neocortex expansion (PubMed:25721503). Promotes the proliferation of basal progenitors by ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3KRB8 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ARHGAP28 | Unsafe / Protein

ENSG00000088756

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); colon / endothelial cells: Medium (Enhanced); colon / peripheral nerve/ganglion: Low (Enhanced); heart muscle / cardiomyocytes: Low (Enhanced); kidney / cells in glomeruli: High (Enhanced); kidney / proximal tubules (cell body): Low (Enhanced); lung ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 38/50 (76.0%); Lung Squamous Cell Carcinoma (validation): 50/68 (73.5%); Testicular Germ Cell Tumor (TCGA): 86/133 (64.7%)

Cancer IHC. testis cancer: 1/10 (10.0%); glioma: 1/11 (9.1%)

Biology. GTPase activator for the Rho-type GTPases by converting them to an inactive GDP-bound state

Curated tissue note. Expressed in testis. Expressed at moderate level in kidney and ovary, and weakly expressed in spleen and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P2N2 | HPA portal (live; not the pinned values)

ARHGEF18 | Unsafe / Protein

ENSG00000104880

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / ciliated cells (cell body): Low (Approved); bronchus / ciliated cells (cilia axoneme): Medium (Approved); bronchus / ciliated cells (ciliary rootlets): Medium (Approved); bronchus / ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Bladder Urothelial Carcinoma (TCGA): 169/169 (100.0%)

Cancer IHC. colorectal cancer: 7/11 (63.6%); lymphoma: 6/11 (54.5%)

Biology. Acts as a guanine nucleotide exchange factor (GEF) for RhoA GTPases. Its activation induces formation of actin stress fibers. Also acts as a GEF for RAC1, inducing production of reactive oxygen species (ROS). Does not act as a GEF for CDC42. The G protein beta-gamma (Gbetagamma) subunits of heterotrimeric G proteins act as activators, explaining the integrated effects of LPA and other G protein-coupled receptor agonists on actin stress ... [full text in table]

Curated tissue note. Expressed in all tissues tested with highest expression in kidney and pancreas. Weakly or not expressed in liver, skeletal muscle and testis. Isoform 1: Expressed in eosinophils (PubMed:29601110). Isoform 2: ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZSZ5 | HPA portal (live; not the pinned values)

ARL17A | Unsafe / Protein

ENSG00000185829

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: High (Uncertain); caudate / neuronal cells: High (Uncertain); cerebellum / cells in molecular layer: High (Uncertain); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%); Breast Invasive Carcinoma (validation): 48/50 (96.0%)

Cancer IHC. melanoma: 8/10 (80.0%); renal cancer: 9/12 (75.0%)

Biology. Putative small GTP-binding protein cycling between an inactive GDP-bound and an active GTP-bound form

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IVW1 | UniProt I3L3L1 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ARL6 | Unsafe / Protein

ENSG00000113966

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Kidney Chromophobe (TCGA): 54/64 (84.4%)

Cancer IHC. liver cancer: 10/12 (83.3%); colorectal cancer: 7/11 (63.6%)

Biology. Involved in membrane protein trafficking at the base of the ciliary organelle. Mediates recruitment onto plasma membrane of the BBSome complex which would constitute a coat complex required for sorting of specific membrane proteins to the primary cilia (PubMed:20603001). Together with BBS1, is necessary for correct trafficking of PKD1 to primary cilia (By similarity). Together with the BBSome complex and LTZL1, controls SMO ciliary ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0F7 | HPA portal (live; not the pinned values)

ARMC12 | Unsafe / RNA

ENSG00000157343

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.8 nTPM (limit 2); basal ganglia: 3.6 nTPM (limit 2); choroid plexus: 5.4 nTPM (limit 2); hippocampal formation: 2.3 nTPM (limit 2); hypothalamus: 2.8 nTPM (limit 2); midbrain: 2.2 nTPM (limit 2); pituitary gland: 10.9 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (validation): 12/58 (20.7%); Glioblastoma Multiforme (TCGA): 19/141 (13.5%); Skin Cuteneous Melanoma (TCGA): 3/99 (3.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Essential for male fertility and sperm mitochondrial sheath formation (By similarity). Required for proper mitochondrial elongation and coiling along the flagellum during the formation of the mitochondrial sheath (By similarity). Facilitates the growth and aggressiveness of neuroblastoma cells (PubMed:30026490). Increases the EZH2 activity and H3K27me3 levels in a RBBP4-dependent manner, and facilitates the enrichment of polycomb ... [full text in table]

Curated tissue note. Expressed in testis (PubMed:33536340, PubMed:35534203). Highly expressed in the mid-piece of the elongated and late spermatids (PubMed:35534203). Expressed at higher levels in neuroblastoma tissues and cell ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T9G4 | HPA portal (live; not the pinned values)

ARMC3 | Unsafe / Protein

ENSG00000165309

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cilia axoneme): Medium (Enhanced); nasopharynx / ciliated cells (tip of cilia): Medium (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 20/50 (40.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 70/176 (39.8%); Ovary Serous Cystadenocarcinoma (validation): 24/81 (29.6%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Essential for male fertility and sperm motility (By similarity). During spermatogenesis, promotes the autophagic degradation of excessive ribosomes, providing energy resources for mitochondria and thus ensuring sperm flagellar motility (PubMed:34428398)

Curated tissue note. Expressed in skeletal muscle, brain, lung, kidney, prostate and testis Mainly expressed in skeletal muscle, liver, spleen and thymus Expressed only in the testis among normal tissues but is expressed ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5W041 | HPA portal (live; not the pinned values)

ARMC8 | Unsafe / Protein

ENSG00000114098

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / ciliated cells (ciliary rootlets): High (Approved); bronchus / goblet cells: High (Approved); caudate / neuronal cells: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. testis cancer: 11/12 (91.7%); colorectal cancer: 9/11 (81.8%)

Biology. Component of the CTLH E3 ubiquitin-protein ligase complex that selectively accepts ubiquitin from UBE2H and mediates ubiquitination and subsequent proteasomal degradation of the transcription factor HBP1

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IUR7 | HPA portal (live; not the pinned values)

ARRDC5 | Unsafe / High RNA

ENSG00000205784

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 20.9 nTPM; pbmc/t-cells; hpa_single_cell_tissue; vital; 38.67 TPM; blood/TREG_NAIVE; dice_treg_naive; vital

Cancer RNA. Glioblastoma Multiforme (validation): 2/58 (3.4%); Uterine Corpus Endometrial Carcinoma (TCGA): 5/176 (2.8%); Bladder Urothelial Carcinoma (TCGA): 3/169 (1.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays an essential role in spermatogenesis. May be involved in the anchoring of the sperm head to the tail during spermatogenesis by affecting SEC22A-mediated SUN5 and NDC1 transport and localization

Curated tissue note. Testis-enriched

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NEK1 | HPA portal (live; not the pinned values)

ART5 | Unsafe / RNA

ENSG00000167311

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.8 nTPM (limit 2); skeletal muscle: 13 nTPM (limit 2); tongue: 10 nTPM (limit 6); choroid plexus: 2.8 nTPM (baseline 2); skeletal muscle: 13 nTPM (baseline 2); tongue: 10 nTPM (baseline 6)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 98/133 (73.7%); Kidney Chromophobe (TCGA): 34/64 (53.1%); Breast Invasive Carcinoma (validation): 7/50 (14.0%)

Cancer IHC. breast cancer: 1/11 (9.1%); ovarian cancer: 1/11 (9.1%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96L15 | HPA portal (live; not the pinned values)

ASB14 | Unsafe / RNA

ENSG00000239388

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. heart muscle: 3.7 nTPM (limit 2); skeletal muscle: 27.1 nTPM (limit 2); tongue: 17.3 nTPM (limit 6); heart muscle: 3.7 nTPM (baseline 2); skeletal muscle: 27.1 nTPM (baseline 2); tongue: 17.3 nTPM (baseline 6)

Cancer RNA. Breast Invasive Carcinoma (validation): 12/50 (24.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 5/176 (2.8%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be a substrate-recognition component of a SCF-like ECS (Elongin-Cullin-SOCS-box protein) E3 ubiquitin-protein ligase complex which mediates the ubiquitination and subsequent proteasomal degradation of target proteins. Plays a role in the inhibition of cardiomyocyte nuclear proliferation by mediating the ubiquitination and degradation of MAPRE2

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NK59 | HPA portal (live; not the pinned values)

ASB3 | Unsafe / Protein

ENSG00000115239

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: High (Uncertain); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. endometrial cancer: 12/12 (100.0%); glioma: 12/12 (100.0%)

Biology. Probable substrate-recognition component of a SCF-like ECS (Elongin-Cullin-SOCS-box protein) E3 ubiquitin-protein ligase complex which mediates the ubiquitination and subsequent proteasomal degradation of target proteins. Recognizes TNFRSF1B (PubMed:15899873). Plays a role in the down-regulation of antiviral innate immunity by targeting MAVS for ubiquitin-proteasomal degradation (PubMed:39266719). Also destabilizes TRAF6 by enhancing ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y575 | HPA portal (live; not the pinned values)

ASB9 | Unsafe / Protein

ENSG00000102048

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / collecting ducts: Low (Enhanced); kidney / distal tubules: Medium (Enhanced); pancreas / pancreatic endocrine cells: High (Enhanced)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 77/99 (77.8%); Rectum Adenocarcinoma (TCGA): 55/88 (62.5%); Lung Adenocarcinoma (validation): 65/105 (61.9%)

Cancer IHC. lymphoma: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. Substrate-recognition component of a cullin-5-RING E3 ubiquitin-protein ligase complex (ECS complex, also named CRL5 complex), which mediates the ubiquitination and subsequent proteasomal degradation of target proteins (PubMed:25654263, PubMed:33268465). The ECS(ASB9) complex catalyzes ubiquitination of creatine kinases CKB and CKMT1A (PubMed:20302626, PubMed:22418839, PubMed:25654263, PubMed:33268465) Does not interact with the ... [full text in table]

Curated tissue note. Predominantly expressed in testis, kidney, and liver

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96DX5 | HPA portal (live; not the pinned values)

ASF1B | Unsafe / Protein

ENSG00000105011

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); colon / glandular cells: Low (Enhanced); duodenum / glandular cells: Medium (Enhanced); heart muscle / cardiomyocytes: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); lymph node / ... [full text in table]

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 283/283 (100.0%); Rectum Adenocarcinoma (TCGA): 88/88 (100.0%); Colon Adenocarcinoma (TCGA): 253/254 (99.6%)

Cancer IHC. testis cancer: 2/12 (16.7%); urothelial cancer: 2/12 (16.7%)

Biology. Histone chaperone that facilitates histone deposition and histone exchange and removal during nucleosome assembly and disassembly (PubMed:11897662, PubMed:14718166, PubMed:15664198, PubMed:16151251, PubMed:21454524, PubMed:26527279). Cooperates with chromatin assembly factor 1 (CAF-1) to promote replication-dependent chromatin assembly (PubMed:11897662, PubMed:14718166, PubMed:15664198, PubMed:16151251). Also involved in the nuclear ... [full text in table]

Curated tissue note. Highly expressed in testis and at lower levels in colon, small intestine and thymus

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NVP2 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ASGR1 | Unsafe / Protein

ENSG00000141505

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. liver / hepatocytes: High (Enhanced); stomach 1 / glandular cells: Low (Enhanced); stomach 2 / glandular cells: Low (Enhanced)

Cancer RNA. Liver Hepatocellular Carcinoma (TCGA): 359/362 (99.2%); Liver Hepatocellular Carcinoma (validation): 224/231 (97.0%); Glioblastoma Multiforme (TCGA): 103/141 (73.0%)

Cancer IHC. liver cancer: 8/12 (66.7%); pancreatic cancer: 1/12 (8.3%)

Biology. Transmembrane protein predominantly expressed on hepatocytes that plays a key role in endocytosis of plasma glycoproteins that lack terminal sialic acid residues. Specifically recognizes terminal galactose and N-acetylgalactosamine residues, facilitating the clearance of desialylated glycoproteins from circulation. Plays thereby a role in a variety of physiological processes, such as removal of desialylated platelets, elimination of ... [full text in table]

Curated tissue note. Expressed exclusively in hepatic parenchymal cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P07306 | HPA portal (live; not the pinned values)

ASPM | Unsafe / RNA

ENSG00000066279

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 10 nTPM (limit 2); thymus: 16.5 nTPM (limit 10); bone marrow: 10 nTPM (baseline 2); thymus: 16.5 nTPM (baseline 10)

Cancer RNA. Colon Adenocarcinoma (validation): 479/486 (98.6%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Rectum Adenocarcinoma (validation): 196/207 (94.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Involved in mitotic spindle regulation and coordination of mitotic processes. The function in regulating microtubule dynamics at spindle poles including spindle orientation, astral microtubule density and poleward microtubule flux seems to depend on the association with the katanin complex formed by KATNA1 and KATNB1. Enhances the microtubule lattice severing activity of KATNA1 by recruiting the katanin complex to microtubules. Can ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZT6 | HPA portal (live; not the pinned values)

ASXL3 | Unsafe / High RNA

ENSG00000141431

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 295.9 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 0.5659 TPM; blood/M2; dice_m2; vital

Cancer RNA. Glioblastoma Multiforme (validation): 45/58 (77.6%); Pancreatic Adenocarcinoma (validation): 4/80 (5.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 4/282 (1.4%)

Cancer IHC. cervical cancer: 12/12 (100.0%); breast cancer: 11/11 (100.0%)

Biology. Putative Polycomb group (PcG) protein. PcG proteins act by forming multiprotein complexes, which are required to maintain the transcriptionally repressive state of homeotic genes throughout development. PcG proteins are not required to initiate repression, but to maintain it during later stages of development. They probably act via methylation of histones, rendering chromatin heritably changed in its expressibility (By similarity). ... [full text in table]

Curated tissue note. Expressed in pancreatic islets, testis, neuroblastoma, head and neck tumor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9C0F0 | HPA portal (live; not the pinned values)

ATAD5 | Unsafe / RNA

ENSG00000176208

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 12.2 nTPM (limit 2); retina: 2.4 nTPM (limit 2); bone marrow: 12.2 nTPM (baseline 2); retina: 2.4 nTPM (baseline 2)

Cancer RNA. Colon Adenocarcinoma (validation): 468/486 (96.3%); Glioblastoma Multiforme (validation): 55/58 (94.8%); Rectum Adenocarcinoma (validation): 188/207 (90.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Has an important role in DNA replication and in maintaining genome integrity during replication stress (PubMed:15983387, PubMed:19755857). Involved in a RAD9A-related damage checkpoint, a pathway that is important in determining whether DNA damage is compatible with cell survival or whether it requires cell elimination by apoptosis (PubMed:15983387). Modulates the RAD9A interaction with BCL2 and thereby induces DNA damage-induced ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96QE3 | HPA portal (live; not the pinned values)

ATG10 | Unsafe / Protein

ENSG00000152348

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 47/50 (94.0%); Rectum Adenocarcinoma (validation): 193/207 (93.2%)

Cancer IHC. colorectal cancer: 9/9 (100.0%); breast cancer: 7/11 (63.6%)

Biology. E2-like enzyme involved in autophagy. Acts as an E2-like enzyme that catalyzes the conjugation of ATG12 to ATG5. ATG12 conjugation to ATG5 is required for autophagy. Likely serves as an ATG5-recognition molecule. Not involved in ATG12 conjugation to ATG3 (By similarity). Plays a role in adenovirus-mediated cell lysis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0Y0 | HPA portal (live; not the pinned values)

ATP1A4 | Unsafe / RNA

ENSG00000132681

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.4 nTPM (limit 2); skeletal muscle: 2.9 nTPM (limit 2); retina: 2.4 nTPM (baseline 2); skeletal muscle: 2.9 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 32/50 (64.0%); Bladder Urothelial Carcinoma (TCGA): 26/169 (15.4%); Breast Invasive Carcinoma (TCGA): 49/1022 (4.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Sperm-specific catalytic alpha subunit of the Na(+)/K(+)-ATPase responsible for ATP-dependent Na(+)/K(+) exchange across the plasma membrane. Transports 3 Na(+) ions out of the cell and 2 K(+) ions into the cell for each ATP hydrolyze (PubMed:16861705). Plays an essential for sperm motility, capacitation, and male fertility by maintaining ionic gradients required for flagellar function and signaling (By similarity)

Curated tissue note. Specifically expressed in testis (PubMed:16175638, PubMed:16861705). Found in very low levels in skeletal muscle. Expressed in mature sperm (at protein level) (PubMed:16175638)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13733 | HPA portal (live; not the pinned values)

ATP6V0C | Unsafe / RNA

ENSG00000185883

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 196.7 nTPM (limit 10); adrenal gland: 239.5 nTPM (limit 2); amygdala: 365.6 nTPM (limit 2); basal ganglia: 389.1 nTPM (limit 2); bone marrow: 180.3 nTPM (limit 2); cerebellum: 459.3 nTPM (limit 2); cerebral cortex: 589.8 nTPM (limit 2); choroid plexus: 220.4 nTPM (limit 2); colon: ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Proton-conducting pore forming subunit of the V0 complex of vacuolar(H+)-ATPase (V-ATPase), a multisubunit enzyme composed of a peripheral complex (V1) that hydrolyzes ATP and a membrane integral complex (V0) that translocates protons (PubMed:33065002, PubMed:36074901). V-ATPase is responsible for acidifying and maintaining the pH of intracellular compartments, and in some cell types, it is targeted to the plasma membrane, where it ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P27449 | HPA portal (live; not the pinned values)

ATP6V1E2 | Unsafe / RNA

ENSG00000250565

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.5 nTPM (limit 2); basal ganglia: 4.2 nTPM (limit 2); hippocampal formation: 3.3 nTPM (limit 2); hypothalamus: 2.6 nTPM (limit 2); midbrain: 3.6 nTPM (limit 2); pituitary gland: 2.7 nTPM (limit 2); retina: 2.3 nTPM (limit 2); spinal cord: 3.8 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (validation): 48/58 (82.8%); Breast Invasive Carcinoma (validation): 40/50 (80.0%); Pancreatic Adenocarcinoma (validation): 29/80 (36.2%)

Cancer IHC. prostate cancer: 1/11 (9.1%); liver cancer: 1/12 (8.3%)

Biology. Subunit of the V1 complex of vacuolar(H+)-ATPase (V-ATPase), a multisubunit enzyme composed of a peripheral complex (V1) that hydrolyzes ATP and a membrane integral complex (V0) that translocates protons. V-ATPase is responsible for acidifying and maintaining the pH of intracellular compartments and in some cell types, is targeted to the plasma membrane, where it is responsible for acidifying the extracellular environment

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96A05 | HPA portal (live; not the pinned values)

ATP8A2 | Unsafe / Protein

ENSG00000132932

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / Purkinje cells: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 18/58 (31.0%); Skin Cuteneous Melanoma (TCGA): 13/99 (13.1%); Prostate Adenocarcinoma (TCGA): 61/480 (12.7%)

Cancer IHC. breast cancer: 3/11 (27.3%); prostate cancer: 3/11 (27.3%)

Biology. Catalytic component of a P4-ATPase flippase complex which catalyzes the hydrolysis of ATP coupled to the transport of aminophospholipids from the outer to the inner leaflet of various membranes and ensures the maintenance of asymmetric distribution of phospholipids (By similarity). Able to translocate phosphatidylserine, but not phosphatidylcholine (PubMed:34403372). Phospholipid translocation also seems to be implicated in vesicle ... [full text in table]

Curated tissue note. Strongly expressed in the brain, cerebellum, retina and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NTI2 | HPA portal (live; not the pinned values)

ATP8B3 | Unsafe / RNA

ENSG00000130270

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 5.8 nTPM (limit 2); bone marrow: 5.8 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 30/50 (60.0%); Kidney Renal Clear Cell Carcinoma (validation): 27/100 (27.0%); Pancreatic Adenocarcinoma (validation): 16/80 (20.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. P4-ATPase flippase which catalyzes the hydrolysis of ATP coupled to the transport of aminophospholipids from the outer to the inner leaflet of various membranes and ensures the maintenance of asymmetric distribution of phospholipids. Phospholipid translocation also seems to be implicated in vesicle formation and in uptake of lipid signaling molecules. May be responsible for the maintenance of asymmetric distribution of ... [full text in table]

Curated tissue note. Isoform 3 was only detected in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O60423 | HPA portal (live; not the pinned values)

ATRNL1 | Unsafe / RNA

ENSG00000107518

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 10.4 nTPM (limit 2); amygdala: 6.3 nTPM (limit 2); basal ganglia: 5 nTPM (limit 2); cerebral cortex: 19.4 nTPM (limit 2); choroid plexus: 4.7 nTPM (limit 2); hippocampal formation: 6.8 nTPM (limit 2); hypothalamus: 4 nTPM (limit 2); kidney: 2.7 nTPM (limit 2); midbrain: 2.4 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 54/58 (93.1%); Breast Invasive Carcinoma (validation): 24/50 (48.0%); Testicular Germ Cell Tumor (TCGA): 45/133 (33.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May play a role in melanocortin signaling pathways that regulate energy homeostasis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VV63 | HPA portal (live; not the pinned values)

AUNIP | Unsafe / RNA

ENSG00000127423

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 6.1 nTPM (limit 2); bone marrow: 6.1 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 109/133 (82.0%); Colon Adenocarcinoma (validation): 385/486 (79.2%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 224/283 (79.2%)

Cancer IHC. melanoma: 9/9 (100.0%); skin cancer: 1/8 (12.5%)

Biology. DNA-binding protein that accumulates at DNA double-strand breaks (DSBs) following DNA damage and promotes DNA resection and homologous recombination (PubMed:29042561). Serves as a sensor of DNA damage: binds DNA with a strong preference for DNA substrates that mimic structures generated at stalled replication forks, and anchors RBBP8/CtIP to DSB sites to promote DNA end resection and ensuing homologous recombination repair ... [full text in table]

Curated tissue note. Expressed in heart, skeletal muscles, placenta and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H7T9 | HPA portal (live; not the pinned values)

AURKA | Unsafe / Protein

ENSG00000087586

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); colon / glandular cells: Low (Enhanced); esophagus / squamous epithelial cells: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); liver / hepatocytes: Low (Enhanced); lung / ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 283/283 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. lymphoma: 5/12 (41.7%); colorectal cancer: 4/12 (33.3%)

Biology. Mitotic serine/threonine kinase that contributes to the regulation of cell cycle progression (PubMed:11039908, PubMed:12390251, PubMed:17125279, PubMed:17360485, PubMed:18615013, PubMed:26246606). Associates with the centrosome and the spindle microtubules during mitosis and plays a critical role in various mitotic events including the establishment of mitotic spindle, centrosome duplication, centrosome separation as well as ... [full text in table]

Curated tissue note. Highly expressed in testis and weakly in skeletal muscle, thymus and spleen. Also highly expressed in colon, ovarian, prostate, neuroblastoma, breast and cervical cancer cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14965 | HPA portal (live; not the pinned values)

AURKC | Unsafe / RNA

ENSG00000105146

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.5 nTPM (limit 2); choroid plexus: 3.6 nTPM (limit 2); liver: 2.6 nTPM (limit 2); spinal cord: 3.3 nTPM (limit 2); cerebral cortex: 2.5 nTPM (baseline 2); choroid plexus: 3.6 nTPM (baseline 2); liver: 2.6 nTPM (baseline 2); spinal cord: 3.3 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 30/50 (60.0%); Glioblastoma Multiforme (validation): 14/58 (24.1%); Testicular Germ Cell Tumor (TCGA): 26/133 (19.5%)

Cancer IHC. liver cancer: 9/12 (75.0%); pancreatic cancer: 7/12 (58.3%)

Biology. Serine/threonine-protein kinase component of the chromosomal passenger complex (CPC), a complex that acts as a key regulator of mitosis. The CPC complex has essential functions at the centromere in ensuring correct chromosome alignment and segregation and is required for chromatin-induced microtubule stabilization and spindle assembly. Also plays a role in meiosis and more particularly in spermatogenesis. Has redundant cellular ... [full text in table]

Curated tissue note. Isoform 1 and isoform 2 are expressed in testis. Elevated expression levels were seen only in a subset of cancer cell lines such as Hep-G2, Huh-7 and HeLa. Expression is maximum at M phase

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UQB9 | HPA portal (live; not the pinned values)

AXDND1 | Unsafe / Protein

ENSG00000162779

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced); nasopharynx / respiratory epithelial cells: Low (Enhanced)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 11/80 (13.8%); Liver Hepatocellular Carcinoma (validation): 4/231 (1.7%); Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May be essential for spermiogenesis and male fertility probably by regulating the manchette dynamics, spermatid head shaping and sperm flagellum assembly

Curated tissue note. Highly expressed in testis. Highly expressed in the round and late spermatids

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T1B0 | HPA portal (live; not the pinned values)

AZI2 | Unsafe / Protein

ENSG00000163512

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); melanoma: 12/12 (100.0%)

Biology. Adapter protein which binds TBK1 and IKBKE playing a role in antiviral innate immunity (PubMed:14560022, PubMed:21931631). Activates serine/threonine-protein kinase TBK1 and facilitates its oligomerization (PubMed:14560022, PubMed:21931631). Enhances the phosphorylation of NF-kappa-B p65 subunit RELA by TBK1 (PubMed:14560022, PubMed:21931631). Promotes TBK1-induced as well as TNF or PMA-induced activation of NF-kappa-B ... [full text in table]

Curated tissue note. Widely expressed (PubMed:14560022). Abundant expression seen in the pancreas and testis (PubMed:14560022)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H6S1 | HPA portal (live; not the pinned values)

AZIN2 | Unsafe / RNA

ENSG00000142920

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.7 nTPM (limit 2); amygdala: 8.5 nTPM (limit 2); basal ganglia: 9.7 nTPM (limit 2); cerebellum: 6.6 nTPM (limit 2); cerebral cortex: 11.8 nTPM (limit 2); choroid plexus: 6.3 nTPM (limit 2); heart muscle: 3.5 nTPM (limit 2); hippocampal formation: 11.2 nTPM (limit 2); hypothalamus: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 49/58 (84.5%); Glioblastoma Multiforme (TCGA): 105/141 (74.5%); Breast Invasive Carcinoma (TCGA): 409/1022 (40.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Antizyme inhibitor (AZI) protein that positively regulates ornithine decarboxylase (ODC) activity and polyamine uptake. AZI is an enzymatically inactive ODC homolog that counteracts the negative effect of ODC antizymes (AZs) OAZ1, OAZ2 and OAZ3 on ODC activity by competing with ODC for antizyme-binding (PubMed:17900240). Inhibits antizyme-dependent ODC degradation and releases ODC monomers from their inactive complex with antizymes, ... [full text in table]

Curated tissue note. Expressed in the neocortex, thalamus, hippocampus, cerebellum, medulla oblongata, gray and white matter. Expressed in neurons, oligodendrocytes, basket, Purkinje and pyramidal cells. Expressed in spermatocytes ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96A70 | HPA portal (live; not the pinned values)

B3GNT5 | Unsafe / Protein

ENSG00000176597

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in molecular layer: Low (Uncertain); cerebellum / Purkinje cells: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. liver cancer: 5/11 (45.5%); prostate cancer: 5/11 (45.5%)

Biology. Beta-1,3-N-acetylglucosaminyltransferase that plays a key role in the synthesis of lacto- or neolacto-series carbohydrate chains on glycolipids, notably by participating in biosynthesis of HNK-1 and Lewis X carbohydrate structures. Has strong activity toward lactosylceramide (LacCer) and neolactotetraosylceramide (nLc(4)Cer; paragloboside), resulting in the synthesis of Lc(3)Cer and neolactopentaosylceramide (nLc(5)Cer), respectively. ... [full text in table]

Curated tissue note. Widely expressed. Highly expressed in lung, colon, placenta, testis, pituitary gland and cerebellum. Weakly expressed in brain, liver, spleen, lymph node and thymus

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BYG0 | HPA portal (live; not the pinned values)

BACH1 | Unsafe / Protein

ENSG00000156273

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Approved); bronchus / ciliated cells (cell body): Low (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: Low (Approved); duodenum / glandular cells: Low (Approved); esophagus / squamous epithelial cells: High (Approved); heart ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. testis cancer: 9/12 (75.0%); cervical cancer: 8/12 (66.7%)

Biology. Transcriptional regulator that acts as a repressor or activator, depending on the context. Binds to NF-E2 DNA binding sites. Plays important roles in coordinating transcription activation and repression by MAFK (By similarity). Together with MAF, represses the transcription of genes under the control of the NFE2L2 oxidative stress pathway (PubMed:24035498)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14867 | HPA portal (live; not the pinned values)

BACH2 | Unsafe / Protein

ENSG00000112182

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / ciliated cells (cell body): Low (Enhanced); bronchus / ciliated cells (ciliary rootlets): Low (Enhanced); bronchus / ciliated cells (tip ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 51/58 (87.9%); Breast Invasive Carcinoma (validation): 15/50 (30.0%); Pancreatic Adenocarcinoma (validation): 10/80 (12.5%)

Cancer IHC. lymphoma: 5/12 (41.7%); testis cancer: 1/11 (9.1%)

Biology. Transcriptional regulator that acts as a repressor or activator (By similarity). Binds to Maf recognition elements (MARE) (By similarity). Plays an important role in coordinating transcription activation and repression by MAFK (By similarity). Induces apoptosis in response to oxidative stress through repression of the antiapoptotic factor HMOX1 (PubMed:17018862). Positively regulates the nuclear import of actin (By similarity). Is a ... [full text in table]

Curated tissue note. B-cell specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BYV9 | HPA portal (live; not the pinned values)

BANF2 | Unsafe / RNA

ENSG00000125888

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pancreas: 19.8 nTPM (limit 4)

Cancer RNA. Liver Hepatocellular Carcinoma (TCGA): 15/362 (4.1%); Pancreatic Adenocarcinoma (TCGA): 5/176 (2.8%); Thyroid Carcinoma (TCGA): 13/495 (2.6%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May play a role in BANF1 regulation and influence tissue-specific roles of BANF1

Curated tissue note. Expressed strongly in testis and pancreas. Also detected in brain, colon, liver, lung, ovary, placenta, prostate, small intestine, spleen and thymus. Not detected in heart, kidney and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H503 | HPA portal (live; not the pinned values)

BARHL2 | Unsafe / RNA

ENSG00000143032

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 43.9 nTPM (limit 2); retina: 2.7 nTPM (limit 2); cerebellum: 43.9 nTPM (baseline 2); retina: 2.7 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%); Ovary Serous Cystadenocarcinoma (TCGA): 2/349 (0.6%); Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Potential regulator of neural basic helix-loop-helix genes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NY43 | HPA portal (live; not the pinned values)

BBS1 | Unsafe / Protein

ENSG00000174483

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); colon / glandular cells: Low (Uncertain); duodenum / glandular cells: Low (Uncertain); heart muscle / cardiomyocytes: Medium (Uncertain); kidney / cells in tubules: Medium (Uncertain); ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Rectum Adenocarcinoma (validation): 205/207 (99.0%); Kidney Renal Clear Cell Carcinoma (validation): 99/100 (99.0%)

Cancer IHC. prostate cancer: 7/11 (63.6%); stomach cancer: 2/12 (16.7%)

Biology. The BBSome complex is thought to function as a coat complex required for sorting of specific membrane proteins to the primary cilia. The BBSome complex is required for ciliogenesis but is dispensable for centriolar satellite function. This ciliogenic function is mediated in part by the Rab8 GDP/GTP exchange factor, which localizes to the basal body and contacts the BBSome. Rab8(GTP) enters the primary cilium and promotes extension of ... [full text in table]

Curated tissue note. Highly expressed in the kidney. Also found in fetal tissue, testis, retina, adipose tissue, heart, skeletal muscle and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NFJ9 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

BCKDHA | Unsafe / Protein

ENSG00000248098

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / glial cells: Medium (Supported); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Together with BCKDHB forms the heterotetrameric E1 subunit of the mitochondrial branched-chain alpha-ketoacid dehydrogenase (BCKD) complex. The BCKD complex catalyzes the multi-step oxidative decarboxylation of alpha-ketoacids derived from the branched-chain amino-acids valine, leucine and isoleucine producing CO2 and acyl-CoA which is subsequently utilized to produce energy. The E1 subunit catalyzes the first step with the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P12694 | HPA portal (live; not the pinned values)

BCL2L14 | Unsafe / Protein

ENSG00000121380

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); caudate / glial cells: Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in granular layer: Medium (Enhanced); cerebral cortex / glial cells: Medium (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 205/207 (99.0%); Colon Adenocarcinoma (validation): 478/486 (98.4%); Colon Adenocarcinoma (TCGA): 223/254 (87.8%)

Cancer IHC. renal cancer: 8/12 (66.7%); glioma: 3/10 (30.0%)

Biology. Plays a role in apoptosis

Curated tissue note. Isoform 1 is widely expressed. Isoform 2 is testis-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BZR8 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

BEAN1 | Unsafe / RNA

ENSG00000166546

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.2 nTPM (limit 2); cerebral cortex: 2.2 nTPM (baseline 2)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 30/80 (37.5%); Pancreatic Adenocarcinoma (TCGA): 55/176 (31.2%); Lung Adenocarcinoma (TCGA): 153/497 (30.8%)

Cancer IHC. liver cancer: 8/12 (66.7%); pancreatic cancer: 6/9 (66.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3B7T3 | HPA portal (live; not the pinned values)

BEND2 | Unsafe / High RNA

ENSG00000177324

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 218.7 nTPM; pbmc/platelets; hpa_single_cell_tissue; vital; 5.442 TPM; blood/basophil; hpa_immune_cell_samples; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Pancreatic Adenocarcinoma (TCGA): 1/176 (0.6%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/9 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NDZ0 | HPA portal (live; not the pinned values)

BEND3 | Unsafe / Protein

ENSG00000178409

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 457/486 (94.0%); Testicular Germ Cell Tumor (TCGA): 119/133 (89.5%); Rectum Adenocarcinoma (validation): 180/207 (87.0%)

Cancer IHC. liver cancer: 11/12 (91.7%); prostate cancer: 9/10 (90.0%)

Biology. Transcriptional repressor which associates with the NoRC (nucleolar remodeling complex) complex and plays a key role in repressing rDNA transcription. The sumoylated form modulates the stability of the NoRC complex component BAZ2A/TIP5 by controlling its USP21-mediated deubiquitination (PubMed:21914818, PubMed:26100909). Binds to unmethylated major satellite DNA and is involved in the recruitment of the Polycomb repressive complex 2 ... [full text in table]

Curated tissue note. Expressed at least in heart, kidney, liver, ovary and spleen, with highest levels in spleen and lowest in heart. Expressed on the surface of T-cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T5X7 | HPA portal (live; not the pinned values)

BEND4 | Unsafe / High RNA

ENSG00000188848

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 31.5 nTPM; eye/muller glia cells; hpa_single_cell_tissue; vital; 41.05 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Prostate Adenocarcinoma (TCGA): 307/480 (64.0%); Testicular Germ Cell Tumor (TCGA): 67/133 (50.4%); Glioblastoma Multiforme (validation): 9/58 (15.5%)

Cancer IHC. breast cancer: 12/12 (100.0%); endometrial cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZU67 | HPA portal (live; not the pinned values)

BFSP1 | Unsafe / Protein

ENSG00000125864

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. eye / lens fiber cells: Medium (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 34/50 (68.0%); Skin Cuteneous Melanoma (TCGA): 41/99 (41.4%); Testicular Germ Cell Tumor (TCGA): 45/133 (33.8%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Required for the correct formation of lens intermediate filaments as part of a complex composed of BFSP1, BFSP2 and CRYAA (PubMed:28935373). Involved in altering the calcium regulation of MIP water permeability (PubMed:30790544)

Curated tissue note. Expressed in the cortex and nucleus of the retina lens (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q12934 | HPA portal (live; not the pinned values)

BIRC5 | Unsafe / Protein

ENSG00000089685

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. 326.2 nTPM; placenta/mixed immune cells; hpa_single_cell_tissue; vital; 26.19 TPM; blood/T-reg; hpa_immune_cell_samples; vital

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 283/283 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%)

Cancer IHC. testis cancer: 9/12 (75.0%); urothelial cancer: 4/12 (33.3%)

Biology. Multitasking protein that has dual roles in promoting cell proliferation and preventing apoptosis (PubMed:20627126, PubMed:21364656, PubMed:25778398, PubMed:28218735, PubMed:9859993). Component of a chromosome passage protein complex (CPC) which is essential for chromosome alignment and segregation during mitosis and cytokinesis (PubMed:16322459). Acts as an important regulator of the localization of this complex; directs CPC movement ... [full text in table]

Curated tissue note. Expressed only in fetal kidney and liver, and to lesser extent, lung and brain (PubMed:10626797). Abundantly expressed in adenocarcinoma (lung, pancreas, colon, breast, and prostate) and in high-grade ... [full text in table]

Therapeutic evidence. Trial registration only.

UniProt O15392 | Trial/source record | HPA portal (live; not the pinned values)

BLOC1S3 | Unsafe / Protein

ENSG00000189114

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / basal cells: Medium (Approved); bronchus / ciliated cells (cell body): Medium (Approved); cerebellum / Purkinje cells: Low (Approved); colon / endothelial cells: Low (Approved); colon / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Colon Adenocarcinoma (validation): 484/486 (99.6%)

Cancer IHC. colorectal cancer: 11/12 (91.7%); lymphoma: 11/12 (91.7%)

Biology. Component of the BLOC-1 complex, a complex that is required for normal biogenesis of lysosome-related organelles (LRO), such as platelet dense granules and melanosomes. In concert with the AP-3 complex, the BLOC-1 complex is required to target membrane protein cargos into vesicles assembled at cell bodies for delivery into neurites and nerve terminals. The BLOC-1 complex, in association with SNARE proteins, is also proposed to be ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6QNY0 | HPA portal (live; not the pinned values)

BLTP1 | Unsafe / Protein

ENSG00000138688

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): Medium (Approved); bronchus / ciliated cells (tip of cilia): Medium (Approved); caudate / neuronal cells: Low (Approved); colon / glandular cells: Low (Approved); duodenum / glandular cells: Medium (Approved); heart muscle / cardiomyocytes: Medium ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. endometrial cancer: 2/12 (16.7%); prostate cancer: 2/12 (16.7%)

Biology. Bridge-like lipid transfer protein that functions as molecular bridges between endoplasmic reticulum and the membranes targeted for lipid delivery (PubMed:35491307, PubMed:40269155). Forms a tunnel with multiple beta-grooves that allows the transport of phospholipids (By similarity). Provides phosphatidylethanolamine for glycosylphosphatidylinositol (GPI) anchor synthesis in the endoplasmic reticulum (PubMed:35015055). Plays a role in ... [full text in table]

Curated tissue note. Highly expressed in testis and ovary. Weakly or not expressed in other tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q2LD37 | HPA portal (live; not the pinned values)

BLZF1 | Unsafe / Protein

ENSG00000117475

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: Medium (Supported); bronchus / respiratory epithelial cells: Medium (Supported); caudate / glial cells: Medium (Supported); caudate / neuronal cells: Medium (Supported); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Required for normal Golgi structure and for protein transport from the endoplasmic reticulum (ER) through the Golgi apparatus to the cell surface

Curated tissue note. Detected in adrenal gland (PubMed:9129147)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H2G9 | HPA portal (live; not the pinned values)

BNC1 | Unsafe / RNA

ENSG00000169594

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 14.6 nTPM (limit 10); esophagus: 12.2 nTPM (limit 6); adipose tissue: 14.6 nTPM (baseline 10); esophagus: 12.2 nTPM (baseline 6)

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 456/492 (92.7%); Lung Squamous Cell Carcinoma (validation): 52/68 (76.5%); Lung Squamous Cell Carcinoma (TCGA): 346/489 (70.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcriptional activator (By similarity). It is likely involved in the regulation of keratinocytes terminal differentiation in squamous epithelia and hair follicles (PubMed:8034748). Required for the maintenance of spermatogenesis (By similarity). It is involved in the positive regulation of oocyte maturation, probably acting through the control of BMP15 levels and regulation of AKT signaling cascade (PubMed:30010909). May also play a ... [full text in table]

Curated tissue note. In epidermis, primarily detected in cells of the basal or immediately suprabasal layers (at protein level) (PubMed:16891417). In hair follicles, mainly expressed in the outer root sheath (at protein level) ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q01954 | HPA portal (live; not the pinned values)

BOD1L2 | Unsafe / High RNA

ENSG00000228075

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 32.2 nTPM; testis/macrophages; hpa_single_cell_tissue; vital; 0.2188 TPM; blood/NK; dice_nk; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. prostate cancer: 12/12 (100.0%); pancreatic cancer: 8/10 (80.0%)

Biology. May play a role in proper chromosome biorientation through the detection or correction of syntelic attachments in mitotic spindles

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYS8 | HPA portal (live; not the pinned values)

BPIFA1 | Unsafe / Protein

ENSG00000198183

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. nasopharynx / respiratory epithelial cells: Medium (Supported)

Cancer RNA. Lung Adenocarcinoma (validation): 74/105 (70.5%); Lung Adenocarcinoma (TCGA): 311/497 (62.6%); Lung Squamous Cell Carcinoma (validation): 35/68 (51.5%)

Cancer IHC. ovarian cancer: 1/11 (9.1%); breast cancer: 0/11 (0.0%)

Biology. Lipid-binding protein which shows high specificity for the surfactant phospholipid dipalmitoylphosphatidylcholine (DPPC) (PubMed:25223608). Plays a role in the innate immune responses of the upper airways (PubMed:23132494, PubMed:23499554). Reduces the surface tension in secretions from airway epithelia and inhibits the formation of biofilm by pathogenic Gram-negative bacteria, such as P.aeruginosa and K.pneumoniae (PubMed:23132494, ... [full text in table]

Curated tissue note. Highly expressed in lung, upper airways and nasopharyngeal regions, including trachea and nasal epithelium (at protein level) (PubMed:11018263, PubMed:11251963, PubMed:11425234, PubMed:12409287, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NP55 | HPA portal (live; not the pinned values)

BPIFA2 | Unsafe / Protein

ENSG00000131050

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. salivary gland / glandular cells: High (Enhanced)

Cancer RNA. Kidney Chromophobe (TCGA): 19/64 (29.7%); Lung Adenocarcinoma (TCGA): 88/497 (17.7%); Lung Adenocarcinoma (validation): 16/105 (15.2%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Has strong antibacterial activity against P.aeruginosa

Curated tissue note. Detected in submandibular gland. Secreted into saliva

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96DR5 | HPA portal (live; not the pinned values)

BPIFC | Unsafe / RNA

ENSG00000184459

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 44.2 nTPM (limit 10); skin: 44.2 nTPM (baseline 10)

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 105/492 (21.3%); Skin Cuteneous Melanoma (TCGA): 19/99 (19.2%); Lung Squamous Cell Carcinoma (TCGA): 9/489 (1.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Detected in the basal layer of the epidermis from inflammatory skin from psoriasis patients, but not in normal skin

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NFQ6 | HPA portal (live; not the pinned values)

BRAP | Unsafe / Protein

ENSG00000089234

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); glioma: 12/12 (100.0%)

Biology. Negatively regulates MAP kinase activation by limiting the formation of Raf/MEK complexes probably by inactivation of the KSR1 scaffold protein. Also acts as a Ras responsive E3 ubiquitin ligase that, on activation of Ras, is modified by auto-polyubiquitination resulting in the release of inhibition of Raf/MEK complex formation. May also act as a cytoplasmic retention protein with a role in regulating nuclear transport

Curated tissue note. Expressed in breast epithelial cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z569 | HPA portal (live; not the pinned values)

BRCA2 | Unsafe / RNA

ENSG00000139618

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 5.4 nTPM (limit 2); bone marrow: 5.4 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Colon Adenocarcinoma (validation): 464/486 (95.5%); Rectum Adenocarcinoma (validation): 195/207 (94.2%)

Cancer IHC. breast cancer: 10/11 (90.9%); colorectal cancer: 8/12 (66.7%)

Biology. Tumor suppressor protein that maintains genome stability primarily by repairing damaged DNA through homologous recombination (HR) (PubMed:11239456, PubMed:12442171, PubMed:15115758, PubMed:15199141, PubMed:15671039, PubMed:15937124, PubMed:17515903, PubMed:17515904, PubMed:18317453, PubMed:19303847, PubMed:20729832, PubMed:20729858, PubMed:20729859, PubMed:21719596, PubMed:27941124, PubMed:37499663, PubMed:37515771). Facilitates the ... [full text in table]

Curated tissue note. Highest levels of expression in breast and thymus, with slightly lower levels in lung, ovary and spleen

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P51587 | HPA portal (live; not the pinned values)

BRIP1 | Unsafe / Protein

ENSG00000136492

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: High (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular layer: High ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 475/486 (97.7%); Rectum Adenocarcinoma (validation): 195/207 (94.2%); Glioblastoma Multiforme (validation): 51/58 (87.9%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); melanoma: 12/12 (100.0%)

Biology. DNA-dependent ATPase and 5'-3' DNA helicase required for the maintenance of chromosomal stability (PubMed:11301010, PubMed:14983014, PubMed:16116421, PubMed:16153896, PubMed:17596542, PubMed:36608669). Acts late in the Fanconi anemia pathway, after FANCD2 ubiquitination (PubMed:14983014, PubMed:16153896). Involved in the repair of DNA double-strand breaks by homologous recombination in a manner that depends on its association with ... [full text in table]

Curated tissue note. Ubiquitously expressed, with highest levels in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BX63 | HPA portal (live; not the pinned values)

BRME1 | Unsafe / Protein

ENSG00000132016

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / neuronal cells: Low (Enhanced); colon / glandular cells: Medium (Enhanced); duodenum / glandular cells: Medium (Enhanced); esophagus / squamous epithelial cells: High (Enhanced); kidney / cells in glomeruli: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); oral mucosa / ... [full text in table]

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 201/283 (71.0%); Glioblastoma Multiforme (validation): 34/58 (58.6%); Rectum Adenocarcinoma (validation): 82/207 (39.6%)

Cancer IHC. urothelial cancer: 6/10 (60.0%); skin cancer: 5/11 (45.5%)

Biology. Meiotic recombination factor component of recombination bridges involved in meiotic double-strand break repair. Modulates the localization of recombinases DMC1:RAD51 to meiotic double-strand break (DSB) sites through the interaction with and stabilization of the BRCA2:HSF2BP complex during meiotic recombination. Indispensable for the DSB repair, homologous synapsis, and crossover formation that are needed for progression past metaphase ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q0VDD7 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

BRWD1 | Unsafe / Protein

ENSG00000185658

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. ovarian cancer: 12/12 (100.0%); colorectal cancer: 11/11 (100.0%)

Biology. May be a transcriptional activator. May be involved in chromatin remodeling (By similarity). Required for regulation of cell morphology and cytoskeletal organization (PubMed:21834987)

Curated tissue note. Ubiquitously expressed (PubMed:12359327). Expressed in respiratory epithelial cells and testis spermatozoa

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NSI6 | HPA portal (live; not the pinned values)

BSPH1 | Unsafe / RNA

ENSG00000188334

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 7.3 nTPM (limit 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Liver Hepatocellular Carcinoma (validation): 1/231 (0.4%); Ovary Serous Cystadenocarcinoma (TCGA): 1/349 (0.3%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Binds sperm in vitro and promotes sperm capacitation. Specifically promotes capacitation induced by high density lipoproteins (HDLs). Also binds heparin, phospholipid liposomes, and weakly to gelatin. Does not bind chondroitin sulfate B

Curated tissue note. Expressed only in the epididymis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q075Z2 | HPA portal (live; not the pinned values)

BTG4 | Unsafe / RNA

ENSG00000137707

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 4.3 nTPM (limit 2); choroid plexus: 4.3 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%); Uterine Corpus Endometrial Carcinoma (TCGA): 3/176 (1.7%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Adapter protein that bridges CNOT7, a catalytic subunit of the CCR4-NOT complex, to EIF4E (By similarity). Facilitates maternal mRNAs decay during the maturation of oocytes and in the fertilized egg, and is required for the maternal-zygotic transition (MZT), zygotic cleavage and initiation of embryonic development (PubMed:32502391)

Curated tissue note. Expressed in oocytes after germinal vesicle breakdown (PubMed:32502391). Expressed in testis and in olfactory epithelium

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NY30 | HPA portal (live; not the pinned values)

BUB1 | Unsafe / RNA

ENSG00000169679

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 11.7 nTPM (limit 2); duodenum: 3.8 nTPM (limit 2); lymph node: 15.6 nTPM (limit 6); small intestine: 3.4 nTPM (limit 2); smooth muscle: 2.1 nTPM (limit 2); thymus: 26.8 nTPM (limit 10); bone marrow: 11.7 nTPM (baseline 2); duodenum: 3.8 nTPM (baseline 2); lymph node: 15.6 nTPM ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 482/486 (99.2%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Rectum Adenocarcinoma (validation): 203/207 (98.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Serine/threonine-protein kinase that performs 2 crucial functions during mitosis: it is essential for spindle-assembly checkpoint signaling and for correct chromosome alignment. Has a key role in the assembly of checkpoint proteins at the kinetochore, being required for the subsequent localization of CENPF, BUB1B, CENPE and MAD2L1. Required for the kinetochore localization of PLK1. Required for centromeric enrichment of AUKRB in ... [full text in table]

Curated tissue note. High expression in testis and thymus, less in colon, spleen, lung and small intestine. Expressed in fetal thymus, bone marrow, heart, liver, spleen and thymus. Expression is associated with cells/tissues with ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43683 | HPA portal (live; not the pinned values)

BUB1B | Unsafe / RNA

ENSG00000156970

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 17.6 nTPM (limit 2); choroid plexus: 12.6 nTPM (limit 2); duodenum: 4.5 nTPM (limit 2); lymph node: 12.3 nTPM (limit 6); small intestine: 4 nTPM (limit 2); thymus: 34.8 nTPM (limit 10); bone marrow: 17.6 nTPM (baseline 2); choroid plexus: 12.6 nTPM (baseline 2); duodenum: 4.5 nTPM ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 132/133 (99.2%); Colon Adenocarcinoma (validation): 482/486 (99.2%); Rectum Adenocarcinoma (validation): 202/207 (97.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Kinase which is a component of the mitotic checkpoint complex (MCC), which inhibits APC/C and delays chromosome segregation until all chromosomes are properly attached to the mitotic spindle (PubMed:27509861, PubMed:31201382). The MCC, which consists of CDC20, MAD2L1/MAD2, BUB1B/BUBR1 and BUB3, is a key player of the spindle assembly checkpoint (PubMed:11702782, PubMed:14706340, PubMed:15020684, PubMed:27509861, PubMed:31201382). The ... [full text in table]

Curated tissue note. Highly expressed in thymus followed by spleen. Preferentially expressed in tissues with a high mitotic index

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O60566 | HPA portal (live; not the pinned values)

C10orf67 | Unsafe / Protein

ENSG00000179133

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): Low (Approved); bronchus / ciliated cells (tip of cilia): Low (Approved); heart muscle / cardiomyocytes: Low (Approved); kidney / cells in tubules: Low (Approved); nasopharynx / ciliated cells (cell body): Low (Approved); nasopharynx / ciliated cells ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 12/133 (9.0%); Bladder Urothelial Carcinoma (TCGA): 8/169 (4.7%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 10/283 (3.5%)

Cancer IHC. testis cancer: 2/10 (20.0%); breast cancer: 1/12 (8.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYJ2 | HPA portal (live; not the pinned values)

C11orf65 | Unsafe / RNA

ENSG00000166323

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. liver: 2.5 nTPM (limit 2); retina: 2.7 nTPM (limit 2); liver: 2.5 nTPM (baseline 2); retina: 2.7 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 13/133 (9.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 15/282 (5.3%); Kidney Chromophobe (TCGA): 3/64 (4.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Acts as an inhibitor of mitochondrial fission. Interacts with MFF and prevents DNM1L recruitment to mitochondria, promoting a more fused mitochondrial network

Curated tissue note. Enriched in the pancreatic beta cell and the testis and is expressed at low levels in other tissues tested

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCR3 | HPA portal (live; not the pinned values)

C11orf71 | Unsafe / RNA

ENSG00000180425

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 10.3 nTPM (limit 2); amygdala: 11.7 nTPM (limit 2); basal ganglia: 13.5 nTPM (limit 2); bone marrow: 3.7 nTPM (limit 2); cerebellum: 7.7 nTPM (limit 2); cerebral cortex: 9.9 nTPM (limit 2); choroid plexus: 7.1 nTPM (limit 2); colon: 6.7 nTPM (limit 6); duodenum: 7.4 nTPM (limit 2); ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 228/231 (98.7%); Thyroid Carcinoma (TCGA): 488/495 (98.6%); Breast Invasive Carcinoma (validation): 49/50 (98.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6IPW1 | HPA portal (live; not the pinned values)

C11orf96 | Unsafe / Protein

ENSG00000187479

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / cells in molecular layer: Medium ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (TCGA): 175/176 (99.4%); Stomach Adenocarcinoma (TCGA): 344/346 (99.4%); Glioblastoma Multiforme (TCGA): 140/141 (99.3%)

Cancer IHC. breast cancer: 11/12 (91.7%); endometrial cancer: 11/12 (91.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z7L8 | HPA portal (live; not the pinned values)

C12orf42 | Unsafe / High RNA

ENSG00000179088

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 69.1 nTPM; lymph node/b-cells; hpa_single_cell_tissue; vital; 34.64 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96LP6 | HPA portal (live; not the pinned values)

C12orf54 | Unsafe / High RNA

ENSG00000177627

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 30.4 nTPM; bronchus/basal respiratory cells; hpa_single_cell_tissue; vital; 9.541 TPM; blood/TREG_MEM; dice_treg_mem; vital

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 16/68 (23.5%); Lung Squamous Cell Carcinoma (TCGA): 46/489 (9.4%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 15/283 (5.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6X4T0 | HPA portal (live; not the pinned values)

C12orf56 | Unsafe / RNA

ENSG00000185306

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.4 nTPM (limit 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (validation): 19/81 (23.5%); Pancreatic Adenocarcinoma (validation): 9/80 (11.2%); Liver Hepatocellular Carcinoma (validation): 19/231 (8.2%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IXR9 | HPA portal (live; not the pinned values)

C12orf60 | Unsafe / RNA

ENSG00000182993

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.6 nTPM (limit 2); heart muscle: 3.2 nTPM (limit 2); kidney: 2.5 nTPM (limit 2); liver: 5.8 nTPM (limit 2); retina: 6.2 nTPM (limit 2); adrenal gland: 4.6 nTPM (baseline 2); heart muscle: 3.2 nTPM (baseline 2); kidney: 2.5 nTPM (baseline 2); liver: 5.8 nTPM (baseline 2); retina: 6.2 ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 44/50 (88.0%); Testicular Germ Cell Tumor (TCGA): 52/133 (39.1%); Liver Hepatocellular Carcinoma (validation): 67/231 (29.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5U649 | HPA portal (live; not the pinned values)

C14orf39 | Unsafe / RNA

ENSG00000179008

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pituitary gland: 4.7 nTPM (limit 2); retina: 2.6 nTPM (limit 2); skeletal muscle: 2.9 nTPM (limit 2); pituitary gland: 4.7 nTPM (baseline 2); retina: 2.6 nTPM (baseline 2); skeletal muscle: 2.9 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 5/58 (8.6%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 16/283 (5.7%); Breast Invasive Carcinoma (validation): 2/50 (4.0%)

Cancer IHC. lung cancer: 11/12 (91.7%); testis cancer: 11/12 (91.7%)

Biology. Meiotic protein that localizes to the central element of the synaptonemal complex and is required for chromosome synapsis during meiotic recombination. Required for the appropriate processing of intermediate recombination nodules before crossover formation

Curated tissue note. Highest expression in retina, skeletal muscle, testis and colon

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N1H7 | HPA portal (live; not the pinned values)

C16orf86 | Unsafe / RNA

ENSG00000159761

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.4 nTPM (limit 2); basal ganglia: 3.9 nTPM (limit 2); choroid plexus: 4.9 nTPM (limit 2); hippocampal formation: 2.3 nTPM (limit 2); hypothalamus: 2.6 nTPM (limit 2); midbrain: 3.2 nTPM (limit 2); pituitary gland: 2.3 nTPM (limit 2); spinal cord: 2.7 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 40/50 (80.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 106/282 (37.6%); Kidney Renal Clear Cell Carcinoma (validation): 16/100 (16.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZW13 | HPA portal (live; not the pinned values)

C16orf87 | Unsafe / Protein

ENSG00000155330

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: Medium (Uncertain); cerebellum / Purkinje cells: Low (Uncertain); cerebral cortex / glial cells: Medium (Uncertain); cerebral cortex / neuropil: Low (Uncertain); esophagus / squamous epithelial cells: Low (Uncertain); heart muscle / cardiomyocytes: Low (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 48/50 (96.0%); Liver Hepatocellular Carcinoma (validation): 219/231 (94.8%)

Cancer IHC. urothelial cancer: 1/10 (10.0%); glioma: 1/12 (8.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PH81 | HPA portal (live; not the pinned values)

C16orf92 | Unsafe / RNA

ENSG00000167194

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 34.7 nTPM (limit 2); retina: 34.7 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (TCGA): 3/141 (2.1%); Lung Adenocarcinoma (validation): 1/105 (1.0%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May play a role in sperm-oocyte fusion during fertilization

Curated tissue note. Testis-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96LL3 | HPA portal (live; not the pinned values)

C16orf95 | Unsafe / Protein

ENSG00000260456

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / endocrine cells: High (Supported); duodenum / endocrine cells: High (Supported); duodenum / glands of Brunner: Medium (Supported); rectum / endocrine cells: High (Supported); small intestine / endocrine cells: High (Supported)

Cancer RNA. Glioblastoma Multiforme (TCGA): 66/141 (46.8%); Liver Hepatocellular Carcinoma (validation): 75/231 (32.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 85/282 (30.1%)

Cancer IHC. stomach cancer: 1/8 (12.5%); pancreatic cancer: 1/10 (10.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H693 | HPA portal (live; not the pinned values)

C17orf67 | Unsafe / Protein

ENSG00000214226

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: Medium (Approved); cerebral cortex / glial cells: Medium (Approved); cerebral cortex / neuropil: Low (Approved); colon / glandular cells: High (Approved); duodenum / glandular cells: Medium (Approved); esophagus / squamous epithelial cells: Medium (Approved); hippocampus / ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 35/50 (70.0%); Liver Hepatocellular Carcinoma (validation): 43/231 (18.6%); Ovary Serous Cystadenocarcinoma (TCGA): 26/349 (7.4%)

Cancer IHC. glioma: 4/10 (40.0%); ovarian cancer: 2/11 (18.2%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q0P5P2 | HPA portal (live; not the pinned values)

C17orf75 | Unsafe / Protein

ENSG00000108666

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: Medium (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / ciliated cells (tip of cilia): Low (Approved); bronchus / goblet cells: High ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. prostate cancer: 11/11 (100.0%); renal cancer: 11/12 (91.7%)

Biology. As component of the WDR11 complex acts together with TBC1D23 to facilitate the golgin-mediated capture of vesicles generated using AP-1 (PubMed:29426865). May have a role in spermatogenesis

Curated tissue note. Highly expressed in testis and also expressed in fetal testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HAS0 | HPA portal (live; not the pinned values)

C18orf54 | Unsafe / RNA

ENSG00000166845

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. spinal cord: 2.4 nTPM (limit 2); spinal cord: 2.4 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 32/50 (64.0%); Testicular Germ Cell Tumor (TCGA): 50/133 (37.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Might play a role in cell proliferation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYD9 | HPA portal (live; not the pinned values)

C19orf18 | Unsafe / Protein

ENSG00000177025

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. thyroid gland / glandular cells: Low (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 50/58 (86.2%); Breast Invasive Carcinoma (validation): 32/50 (64.0%); Glioblastoma Multiforme (TCGA): 28/141 (19.9%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEA5 | HPA portal (live; not the pinned values)

C19orf25 | Unsafe / Protein

ENSG00000119559

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: Medium (Approved); duodenum / glandular cells: Medium (Approved); esophagus / squamous epithelial cells: Medium (Approved); heart muscle / cardiomyocytes: Low (Approved); ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 495/495 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%)

Cancer IHC. skin cancer: 5/10 (50.0%); cervical cancer: 4/12 (33.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UFG5 | HPA portal (live; not the pinned values)

C19orf84 | Unsafe / Protein

ENSG00000262874

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebral cortex / endothelial cells: Low (Uncertain); cerebral cortex / neuronal cells: Medium ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 62/133 (46.6%); Ovary Serous Cystadenocarcinoma (TCGA): 35/349 (10.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 9/176 (5.1%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/9 (0.0%)

Biology. Protein adapter involved in piRNA-directed transposon methylation by connecting PIWIL4-piRNA and DNA methylation machineries. The PIWIL4-piRNA pathway plays a central role during spermatogenesis by directing transposon DNA methylation and silencing, thereby preventing their mobilization, which is essential for the germline integrity

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt I3L1E1 | HPA portal (live; not the pinned values)

C1orf105 | Unsafe / RNA

ENSG00000180999

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. heart muscle: 16.8 nTPM (limit 2); heart muscle: 16.8 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 3/58 (5.2%); Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%); Rectum Adenocarcinoma (validation): 5/207 (2.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95561 | HPA portal (live; not the pinned values)

C1orf141 | Unsafe / RNA

ENSG00000203963

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.7 nTPM (limit 2); retina: 3.1 nTPM (limit 2); choroid plexus: 2.7 nTPM (baseline 2); retina: 3.1 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%); Lung Adenocarcinoma (TCGA): 6/497 (1.2%); Bladder Urothelial Carcinoma (TCGA): 2/169 (1.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JVX7 | HPA portal (live; not the pinned values)

C1orf167 | Unsafe / High RNA

ENSG00000215910

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 95 nTPM; lung/alveolar cells type 1; hpa_single_cell_tissue; vital; 2.9 TPM; blood/Plasmablast; hpa_immune_monaco; vital

Cancer RNA. Ovary Serous Cystadenocarcinoma (validation): 5/81 (6.2%); Pancreatic Adenocarcinoma (validation): 4/80 (5.0%); Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%)

Cancer IHC. prostate cancer: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5SNV9 | HPA portal (live; not the pinned values)

C1orf226 | Unsafe / Protein

ENSG00000239887

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / cells in molecular layer: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 483/486 (99.4%); Rectum Adenocarcinoma (validation): 204/207 (98.6%); Breast Invasive Carcinoma (validation): 49/50 (98.0%)

Cancer IHC. melanoma: 12/12 (100.0%); ovarian cancer: 11/11 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A1L170 | HPA portal (live; not the pinned values)

C1orf56 | Unsafe / Protein

ENSG00000143443

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / proximal tubules (cell body): Medium (Enhanced)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%); Thyroid Carcinoma (TCGA): 490/495 (99.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Involved in control of cellular proliferation. Onconcogenic modifier contributing to the tumor suppressor function of DNMT3B

Curated tissue note. Plasma. Overexpressed in lymphomas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BUN1 | HPA portal (live; not the pinned values)

C22orf23 | Unsafe / RNA

ENSG00000128346

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.5 nTPM (limit 2); basal ganglia: 2.2 nTPM (limit 2); bone marrow: 3 nTPM (limit 2); cerebral cortex: 7.2 nTPM (limit 2); choroid plexus: 10.8 nTPM (limit 2); duodenum: 2.8 nTPM (limit 2); esophagus: 13.5 nTPM (limit 6); heart muscle: 5 nTPM (limit 2); hippocampal formation: 2.2 ... [full text in table]

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 469/492 (95.3%); Lung Squamous Cell Carcinoma (validation): 63/68 (92.6%); Glioblastoma Multiforme (TCGA): 128/141 (90.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BZE7 | HPA portal (live; not the pinned values)

C22orf42 | Unsafe / Protein

ENSG00000205856

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 26/480 (5.4%); Pancreatic Adenocarcinoma (TCGA): 6/176 (3.4%); Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%)

Cancer IHC. colorectal cancer: 1/11 (9.1%); ovarian cancer: 1/11 (9.1%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6IC83 | HPA portal (live; not the pinned values)

C2CD6 | Unsafe / High RNA

ENSG00000155754

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 32.2 nTPM; brain/oligodendrocytes; hpa_single_cell_tissue; vital; 3.329 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 40/133 (30.1%); Lung Adenocarcinoma (validation): 17/105 (16.2%); Skin Cuteneous Melanoma (TCGA): 7/99 (7.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Auxiliary component of the CatSper complex, a complex involved in sperm cell hyperactivation. Sperm cell hyperactivation is needed for sperm motility which is essential late in the preparation of sperm for fertilization. Required for CatSper complex targeting and trafficking into the quadrilinear nanodomains. Targets the preassembled CatSper complexes to elongating flagella, where it links the channel-carrying vesicles and motor ... [full text in table]

Curated tissue note. Expressed in testis (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q53TS8 | HPA portal (live; not the pinned values)

C4orf46 | Unsafe / Protein

ENSG00000205208

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Uncertain); cerebral cortex / endothelial cells: Low (Uncertain); colon / glandular cells: Low (Uncertain); heart muscle / cardiomyocytes: Medium (Uncertain); kidney / cells in tubules: High (Uncertain); lymph node / non-germinal center cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 450/486 (92.6%); Breast Invasive Carcinoma (validation): 45/50 (90.0%)

Cancer IHC. renal cancer: 4/12 (33.3%); breast cancer: 0/12 (0.0%)

Biology. Expressed in the kidney, in epithelial cells in both proximal tubules and distal convoluted tubules

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q504U0 | HPA portal (live; not the pinned values)

C5orf24 | Unsafe / Protein

ENSG00000181904

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: High (Approved); ... [full text in table]

Cancer RNA. Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. glioma: 12/12 (100.0%); ovarian cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z6I8 | HPA portal (live; not the pinned values)

C5orf34 | Unsafe / Protein

ENSG00000172244

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular layer: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Breast Invasive Carcinoma (validation): 44/50 (88.0%); Lung Squamous Cell Carcinoma (validation): 21/68 (30.9%)

Cancer IHC. breast cancer: 11/11 (100.0%); prostate cancer: 8/11 (72.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96MH7 | HPA portal (live; not the pinned values)

C5orf58 | Unsafe / RNA

ENSG00000234511

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 6.6 nTPM (limit 2); lung: 4.8 nTPM (limit 2); midbrain: 2.3 nTPM (limit 2); spinal cord: 7.5 nTPM (limit 2); bone marrow: 6.6 nTPM (baseline 2); lung: 4.8 nTPM (baseline 2); midbrain: 2.3 nTPM (baseline 2); spinal cord: 7.5 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 65/133 (48.9%); Liver Hepatocellular Carcinoma (validation): 53/231 (22.9%); Breast Invasive Carcinoma (validation): 11/50 (22.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt C9J3I9 | HPA portal (live; not the pinned values)

C5orf63 | Unsafe / RNA

ENSG00000164241

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.9 nTPM (limit 2); amygdala: 2.2 nTPM (limit 2); basal ganglia: 2.5 nTPM (limit 2); bone marrow: 3.5 nTPM (limit 2); choroid plexus: 4.7 nTPM (limit 2); colon: 6.7 nTPM (limit 6); duodenum: 5.5 nTPM (limit 2); heart muscle: 6.5 nTPM (limit 2); hypothalamus: 3.1 nTPM (limit 2); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 190/207 (91.8%); Colon Adenocarcinoma (validation): 446/486 (91.8%); Lung Squamous Cell Carcinoma (validation): 58/68 (85.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NC05 | HPA portal (live; not the pinned values)

C6orf52 | Unsafe / RNA

ENSG00000137434

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 4.3 nTPM (limit 2); basal ganglia: 3.2 nTPM (limit 2); cerebral cortex: 3.3 nTPM (limit 2); choroid plexus: 2.5 nTPM (limit 2); hippocampal formation: 3.7 nTPM (limit 2); hypothalamus: 4.3 nTPM (limit 2); kidney: 4.3 nTPM (limit 2); midbrain: 3.9 nTPM (limit 2); pancreas: 2.2 nTPM (limit ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 41/50 (82.0%); Glioblastoma Multiforme (validation): 42/58 (72.4%); Ovary Serous Cystadenocarcinoma (TCGA): 106/349 (30.4%)

Cancer IHC. breast cancer: 8/11 (72.7%); testis cancer: 8/11 (72.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T4I8 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

C7orf25 | Unsafe / Protein

ENSG00000136197

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 50/50 (100.0%); Liver Hepatocellular Carcinoma (validation): 223/231 (96.5%); Glioblastoma Multiforme (validation): 23/58 (39.7%)

Cancer IHC. lymphoma: 11/12 (91.7%); skin cancer: 10/11 (90.9%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BPX7 | HPA portal (live; not the pinned values)

C7orf57 | Unsafe / Protein

ENSG00000164746

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Low (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); caudate / glial cells: Medium (Enhanced); nasopharynx / ciliated cells (cell body): Low (Enhanced); nasopharynx / ciliated ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 23/58 (39.7%); Kidney Chromophobe (TCGA): 21/64 (32.8%); Testicular Germ Cell Tumor (TCGA): 31/133 (23.3%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEG2 | HPA portal (live; not the pinned values)

C9orf153 | Unsafe / RNA

ENSG00000187753

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. bone marrow: 2.4 nTPM (limit 2); bone marrow: 2.4 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 16/207 (7.7%); Uterine Corpus Endometrial Carcinoma (TCGA): 6/176 (3.4%); Colon Adenocarcinoma (validation): 15/486 (3.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5TBE3 | HPA portal (live; not the pinned values)

C9orf85 | Unsafe / Protein

ENSG00000155621

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bronchus / ciliated cells (cell body): Low (Approved); caudate / glial cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebral cortex / neuronal cells: Medium (Approved); cerebral cortex / neuropil: Medium (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. stomach cancer: 12/12 (100.0%); colorectal cancer: 11/12 (91.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96MD7 | HPA portal (live; not the pinned values)

CABLES2 | Unsafe / RNA

ENSG00000149679

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.9 nTPM (limit 2); amygdala: 4.2 nTPM (limit 2); basal ganglia: 3.9 nTPM (limit 2); bone marrow: 8 nTPM (limit 2); cerebellum: 13.8 nTPM (limit 2); cerebral cortex: 8.6 nTPM (limit 2); choroid plexus: 2.1 nTPM (limit 2); duodenum: 2.8 nTPM (limit 2); heart muscle: 10.2 nTPM (limit ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 476/486 (97.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Unknown. Probably involved in G1-S cell cycle transition

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BTV7 | HPA portal (live; not the pinned values)

CABP2 | Unsafe / RNA

ENSG00000167791

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 7.1 nTPM (limit 2); retina: 7.1 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Required for sound encoding at inner hair cells (IHCs) synapses, likely via inhibition of the inactivation of voltage-gated calcium channel of type 1.3 (Cav1.3) in the IHCs (PubMed:28183797). Required for the normal transfer of light signals through the retina (By similarity)

Curated tissue note. Retina

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NPB3 | HPA portal (live; not the pinned values)

CABYR | Unsafe / RNA

ENSG00000154040

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 5.1 nTPM (limit 2); basal ganglia: 3 nTPM (limit 2); choroid plexus: 12.7 nTPM (limit 2); hippocampal formation: 7 nTPM (limit 2); hypothalamus: 5.3 nTPM (limit 2); midbrain: 5.2 nTPM (limit 2); pituitary gland: 8 nTPM (limit 2); retina: 5.4 nTPM (limit 2); spinal cord: 6.7 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 47/50 (94.0%); Testicular Germ Cell Tumor (TCGA): 124/133 (93.2%); Breast Invasive Carcinoma (TCGA): 843/1022 (82.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May function as a regulator of both motility- and head-associated functions such as capacitation and the acrosome reaction. Isoform 1 binds calcium in vitro. Isoform 2 and isoform 6 probably bind calcium. Isoform 3 and isoform 5 do not bind calcium in vitro. Isoform 4 probably does not bind calcium

Curated tissue note. Expressed in elongating spermatids and spermatozoa (at protein level). Isoform 1 is expressed in testis. Isoform 3 and isoform 5 are also expressed in brain, pancreas and numerous brain tumors

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75952 | HPA portal (live; not the pinned values)

CACNA2D3 | Unsafe / RNA

ENSG00000157445

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4 nTPM (limit 2); amygdala: 3.2 nTPM (limit 2); basal ganglia: 16.2 nTPM (limit 2); cerebellum: 2.2 nTPM (limit 2); cerebral cortex: 16 nTPM (limit 2); heart muscle: 2.5 nTPM (limit 2); hippocampal formation: 7.6 nTPM (limit 2); hypothalamus: 6.8 nTPM (limit 2); midbrain: 2.3 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 24/58 (41.4%); Ovary Serous Cystadenocarcinoma (validation): 10/81 (12.3%); Testicular Germ Cell Tumor (TCGA): 8/133 (6.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. The alpha-2/delta subunit of voltage-dependent calcium channels regulates calcium current density and activation/inactivation kinetics of the calcium channel. Acts as a regulatory subunit for P/Q-type calcium channel (CACNA1A), N-type (CACNA1B), L-type (CACNA1C OR CACNA1D) but not T-type (CACNA1G) (By similarity)

Curated tissue note. Only detected in brain. Not present in lung, testis, aorta, spleen, jejunum, ventricular muscle and kidney (at protein level). According to PubMed:11687876, it is brain-specific, while according to ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZS8 | HPA portal (live; not the pinned values)

CACNA2D4 | Unsafe / RNA

ENSG00000151062

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 19.5 nTPM (limit 2); retina: 19.5 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Pancreatic Adenocarcinoma (validation): 34/80 (42.5%); Glioblastoma Multiforme (validation): 6/58 (10.3%)

Cancer IHC. colorectal cancer: 11/12 (91.7%); endometrial cancer: 10/12 (83.3%)

Biology. The alpha-2/delta subunit of voltage-dependent calcium channels regulates calcium current density and activation/inactivation kinetics of the calcium channel

Curated tissue note. Predominantly expressed in certain types of endocrine cells. Present in the Paneth cells of the small intestine. Also present in the erythroblasts in the fetal liver, in the cells of the zona reticularis of ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z3S7 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CALHM4 | Unsafe / RNA

ENSG00000164451

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3.2 nTPM (limit 2); choroid plexus: 3.2 nTPM (baseline 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 2/349 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Thyroid Carcinoma (TCGA): 2/495 (0.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May assemble to form gap junction channel-like structures involved in intercellular communication. Channel gating and ion conductance are likely regulated by membrane lipids rather than by membrane depolarization or extracellular calcium levels

Curated tissue note. Placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JW98 | HPA portal (live; not the pinned values)

CALN1 | Unsafe / Protein

ENSG00000183166

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 13/58 (22.4%); Testicular Germ Cell Tumor (TCGA): 12/133 (9.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 12/176 (6.8%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Negatively regulates Golgi-to-plasma membrane trafficking by interacting with PI4KB and inhibiting its activity. May play a role in the physiology of neurons and is potentially important in memory and learning

Curated tissue note. Brain specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BXU9 | HPA portal (live; not the pinned values)

CAMK4 | Unsafe / Protein

ENSG00000152495

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: High (Approved); cerebellum / cells in granular layer: High (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 31/58 (53.4%); Breast Invasive Carcinoma (validation): 18/50 (36.0%); Pancreatic Adenocarcinoma (validation): 28/80 (35.0%)

Cancer IHC. lymphoma: 5/12 (41.7%); liver cancer: 3/11 (27.3%)

Biology. Calcium/calmodulin-dependent protein kinase that operates in the calcium-triggered CaMKK-CaMK4 signaling cascade and regulates, mainly by phosphorylation, the activity of several transcription activators, such as CREB1, MEF2D, JUN and RORA, which play pivotal roles in immune response, inflammation, and memory consolidation. In the thymus, regulates the CD4(+)/CD8(+) double positive thymocytes selection threshold during T-cell ontogeny. ... [full text in table]

Curated tissue note. Expressed in brain, thymus, CD4 T-cells, testis and epithelial ovarian cancer tissue

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q16566 | HPA portal (live; not the pinned values)

CAPN6 | Unsafe / RNA

ENSG00000077274

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 11.2 nTPM (limit 2); pancreas: 7.2 nTPM (limit 2); rectum: 8.3 nTPM (limit 6); salivary gland: 13.7 nTPM (limit 6); skeletal muscle: 2.9 nTPM (limit 2); smooth muscle: 7.9 nTPM (limit 2); stomach: 12.6 nTPM (limit 6); tongue: 8.6 nTPM (limit 6); kidney: 11.2 nTPM (baseline 2); pancreas: 7.2 ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 30/50 (60.0%); Pancreatic Adenocarcinoma (TCGA): 100/176 (56.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 142/282 (50.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Microtubule-stabilizing protein that may be involved in the regulation of microtubule dynamics and cytoskeletal organization. May act as a regulator of RAC1 activity through interaction with ARHGEF2 to control lamellipodial formation and cell mobility. Does not seem to have protease activity as it has lost the active site residues (By similarity)

Curated tissue note. Expressed only in placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y6Q1 | HPA portal (live; not the pinned values)

CAPSL | Unsafe / Protein

ENSG00000152611

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (ciliary rootlets): Low (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cell body): Medium ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 75/176 (42.6%); Breast Invasive Carcinoma (validation): 17/50 (34.0%); Ovary Serous Cystadenocarcinoma (validation): 22/81 (27.2%)

Cancer IHC. melanoma: 6/11 (54.5%); stomach cancer: 5/11 (45.5%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WWF8 | HPA portal (live; not the pinned values)

CARF | Unsafe / Protein

ENSG00000138380

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 206/207 (99.5%); Colon Adenocarcinoma (validation): 475/486 (97.7%)

Cancer IHC. breast cancer: 12/12 (100.0%); skin cancer: 12/12 (100.0%)

Biology. Acts as a transcriptional activator that mediates the calcium- and neuron-selective induction of BDNF exon III transcription. Binds to the consensus calcium-response element CaRE1 5'-CTATTTCGAG-3' sequence

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N187 | HPA portal (live; not the pinned values)

CATSPER1 | Unsafe / High RNA

ENSG00000175294

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 101.8 nTPM; lung/granulocytes; hpa_single_cell_tissue; vital; 32.81 TPM; blood/NK; dice_nk; vital

Cancer RNA. Pancreatic Adenocarcinoma (validation): 12/80 (15.0%); Breast Invasive Carcinoma (validation): 1/50 (2.0%); Lung Adenocarcinoma (validation): 2/105 (1.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Pore-forming subunit of the CatSper complex, a sperm-specific voltage-gated calcium channel that plays a central role in calcium-dependent physiological responses essential for successful fertilization, such as sperm hyperactivation, acrosome reaction and chemotaxis towards the oocyte

Curated tissue note. Testis-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEC5 | HPA portal (live; not the pinned values)

CATSPER3 | Unsafe / RNA

ENSG00000152705

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.6 nTPM (limit 2); skeletal muscle: 2.9 nTPM (limit 2); retina: 2.6 nTPM (baseline 2); skeletal muscle: 2.9 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 27/50 (54.0%); Testicular Germ Cell Tumor (TCGA): 13/133 (9.8%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 8/283 (2.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Pore-forming subunit of the CatSper complex, a sperm-specific voltage-gated calcium channel that plays a central role in calcium-dependent physiological responses essential for successful fertilization, such as sperm hyperactivation, acrosome reaction and chemotaxis towards the oocyte

Curated tissue note. Testis-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86XQ3 | HPA portal (live; not the pinned values)

CATSPERB | Unsafe / RNA

ENSG00000133962

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pancreas: 10.4 nTPM (limit 2); pancreas: 10.4 nTPM (baseline 2)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 43/80 (53.8%); Breast Invasive Carcinoma (validation): 26/50 (52.0%); Colon Adenocarcinoma (validation): 55/486 (11.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Auxiliary component of the CatSper complex, a complex involved in sperm cell hyperactivation. Sperm cell hyperactivation is needed for sperm motility which is essential late in the preparation of sperm for fertilization

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H7T0 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CATSPERD | Unsafe / High RNA

ENSG00000174898

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 177.1 nTPM; lung/ciliated cells; hpa_single_cell_tissue; vital; 0.6812 TPM; blood/TH1; dice_th1; vital

Cancer RNA. Pancreatic Adenocarcinoma (validation): 2/80 (2.5%); Ovary Serous Cystadenocarcinoma (validation): 2/81 (2.5%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. prostate cancer: 1/11 (9.1%); breast cancer: 0/11 (0.0%)

Biology. Auxiliary component of the CatSper complex, a complex involved in sperm cell hyperactivation. Sperm cell hyperactivation is needed for sperm motility which is essential late in the preparation of sperm for fertilization. Required for CATSPER1 stability before intraflagellar transport and/or incorporation of the CatSper complex channel into the flagellar membrane

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86XM0 | HPA portal (live; not the pinned values)

CATSPERE | Unsafe / High RNA

ENSG00000179397

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 40 nTPM; brain/oligodendrocytes; hpa_single_cell_tissue; vital; 6.076 TPM; blood/CD8_NAIVE; dice_cd8_naive; vital

Cancer RNA. Glioblastoma Multiforme (validation): 13/58 (22.4%); Kidney Renal Papillary Cell Carcinoma (TCGA): 2/282 (0.7%); Glioblastoma Multiforme (TCGA): 1/141 (0.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Auxiliary component of the CatSper complex, a complex involved in sperm cell hyperactivation. Sperm cell hyperactivation is needed for sperm motility which is essential late in the preparation of sperm for fertilization

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5SY80 | HPA portal (live; not the pinned values)

CATSPERG | Unsafe / RNA

ENSG00000099338

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. cerebellum: 2.4 nTPM (limit 2); cerebellum: 2.4 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 15/50 (30.0%); Pancreatic Adenocarcinoma (validation): 16/80 (20.0%); Ovary Serous Cystadenocarcinoma (validation): 13/81 (16.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Auxiliary component of the CatSper complex, a complex involved in sperm cell hyperactivation. Sperm cell hyperactivation is needed for sperm motility which is essential late in the preparation of sperm for fertilization

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZRH7 | HPA portal (live; not the pinned values)

CATSPERZ | Unsafe / RNA

ENSG00000219435

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.1 nTPM (limit 2); basal ganglia: 2.7 nTPM (limit 2); cerebral cortex: 2.9 nTPM (limit 2); hippocampal formation: 3.3 nTPM (limit 2); amygdala: 3.1 nTPM (baseline 2); basal ganglia: 2.7 nTPM (baseline 2); cerebral cortex: 2.9 nTPM (baseline 2); hippocampal formation: 3.3 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 10/133 (7.5%); Skin Cuteneous Melanoma (TCGA): 7/99 (7.1%); Kidney Renal Clear Cell Carcinoma (TCGA): 29/521 (5.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Auxiliary component of the CatSper complex, a complex involved in sperm cell hyperactivation. Sperm cell hyperactivation is needed for sperm motility which is essential late in the preparation of sperm for fertilization. Required for a distribution of the CatSper complex in linear quadrilateral nanodomains along the flagellum, maximizing fertilization inside the mammalian female reproductive tract. Together with EFCAB9, associates with ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NTU4 | HPA portal (live; not the pinned values)

CBLL1 | Unsafe / Protein

ENSG00000105879

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Enhanced); adrenal gland / glandular cells: High (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); endometrial cancer: 12/12 (100.0%)

Biology. E3 ubiquitin-protein ligase that mediates ubiquitination of several tyrosine-phosphorylated Src substrates, including CDH1, CTTN and DOK1 (By similarity). Targets CDH1 for endocytosis and degradation (By similarity). Associated component of the WMM complex, a complex that mediates N6-methyladenosine (m6A) methylation of RNAs, a modification that plays a role in the efficiency of mRNA splicing and RNA processing (PubMed:29507755). Its ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q75N03 | HPA portal (live; not the pinned values)

CBLN1 | Unsafe / RNA

ENSG00000102924

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 11.4 nTPM (limit 10); basal ganglia: 3.9 nTPM (limit 2); cerebellum: 439.8 nTPM (limit 2); cerebral cortex: 5.5 nTPM (limit 2); heart muscle: 4.4 nTPM (limit 2); hypothalamus: 37.4 nTPM (limit 2); midbrain: 17.7 nTPM (limit 2); pancreas: 2.7 nTPM (limit 2); pituitary gland: 4.1 nTPM ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 254/495 (51.3%); Testicular Germ Cell Tumor (TCGA): 63/133 (47.4%); Glioblastoma Multiforme (validation): 17/58 (29.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Required for synapse integrity and synaptic plasticity. During cerebellar synapse formation, essential for the matching and maintenance of pre- and post-synaptic elements at parallel fiber-Purkinje cell synapses, the establishment of the proper pattern of climbing fiber-Purkinje cell innervation, and induction of long-term depression at parallel fiber-Purkinje cell synapses. Plays a role as a synaptic organizer that acts ... [full text in table]

Curated tissue note. In the Purkinje cells postsynaptic structures. In the cerebellum, cerebellin is much less abundant than [des-Ser1]-cerebellin

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P23435 | HPA portal (live; not the pinned values)

CBX2 | Unsafe / Protein

ENSG00000173894

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); colon / glandular cells: Low (Approved); duodenum / glandular cells: Low (Approved); esophagus / squamous epithelial cells: Low (Approved); lung / alveolar cells: Low (Approved); lymph node / ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 129/176 (73.3%)

Cancer IHC. glioma: 6/12 (50.0%); testis cancer: 4/10 (40.0%)

Biology. Component of a Polycomb group (PcG) multiprotein PRC1-like complex, a complex class required to maintain the transcriptionally repressive state of many genes, including Hox genes, throughout development (PubMed:21282530). PcG PRC1 complex acts via chromatin remodeling and modification of histones; it mediates monoubiquitination of histone H2A 'Lys-119', rendering chromatin heritably changed in its expressibility (PubMed:21282530). ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14781 | HPA portal (live; not the pinned values)

CBX5 | Unsafe / Protein

ENSG00000094916

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. cervical cancer: 11/11 (100.0%); prostate cancer: 9/9 (100.0%)

Biology. Component of heterochromatin that recognizes and binds histone H3 tails methylated at 'Lys-9' (H3K9me), leading to epigenetic repression (PubMed:40440427). In contrast, it is excluded from chromatin when 'Tyr-41' of histone H3 is phosphorylated (H3Y41ph) (PubMed:19783980). Also recognizes and binds histone H1.4 methylated at 'Lys-26' (H1.4K26me) (PubMed:16127177). Excluded from chromatin when histone H1.4 is Simultaneously methylated ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P45973 | HPA portal (live; not the pinned values)

CBY3 | Unsafe / RNA

ENSG00000204659

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. midbrain: 2.8 nTPM (limit 2); spinal cord: 3 nTPM (limit 2); midbrain: 2.8 nTPM (baseline 2); spinal cord: 3 nTPM (baseline 2)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Bladder Urothelial Carcinoma (TCGA): 2/169 (1.2%); Skin Cuteneous Melanoma (TCGA): 1/99 (1.0%)

Cancer IHC. glioma: 12/12 (100.0%); ovarian cancer: 12/12 (100.0%)

Biology. Plays a key role in the correct positioning of the annulus, a septin-based ring structure in the sperm flagellum, serving both as a physical barrier and a membrane diffusion barrier that separates the midpiece (MP) from the principal piece (PP) (By similarity). This positioning is essential for proper sperm motility and function (By similarity). CBY3 interacts with CIBAR1 to form a complex which localizes to the curved membrane region ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NI87 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CCDC110 | Unsafe / RNA

ENSG00000168491

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 2.4 nTPM (limit 2); pancreas: 29.7 nTPM (limit 2); pituitary gland: 2.8 nTPM (limit 2); retina: 7.7 nTPM (limit 2); kidney: 2.4 nTPM (baseline 2); pancreas: 29.7 nTPM (baseline 2); pituitary gland: 2.8 nTPM (baseline 2); retina: 7.7 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 39/50 (78.0%); Kidney Renal Clear Cell Carcinoma (validation): 42/100 (42.0%); Prostate Adenocarcinoma (TCGA): 159/480 (33.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Expressed specifically in testis. Also expressed in tumors of different origins

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TBZ0 | HPA portal (live; not the pinned values)

CCDC122 | Unsafe / Protein

ENSG00000151773

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Medium (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / endothelial cells: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Pancreatic Adenocarcinoma (validation): 58/80 (72.5%); Thyroid Carcinoma (TCGA): 277/495 (56.0%)

Cancer IHC. cervical cancer: 9/10 (90.0%); skin cancer: 10/12 (83.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T0U0 | HPA portal (live; not the pinned values)

CCDC136 | Unsafe / Protein

ENSG00000128596

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Medium (Enhanced); cerebellum / molecular layer cells - cytoplasm/membrane: High (Enhanced); cerebellum / processes in molecular layer: Medium (Enhanced); cerebellum / Purkinje cells - cytoplasm/membrane: Medium (Enhanced); cerebellum / synaptic glomeruli - core: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 121/141 (85.8%); Testicular Germ Cell Tumor (TCGA): 93/133 (69.9%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May play a role in acrosome formation in spermatogenesis and in fertilization

Curated tissue note. Expressed in gastric tissues. Down-regulated in gastric cancer

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96JN2 | HPA portal (live; not the pinned values)

CCDC138 | Unsafe / Protein

ENSG00000163006

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); colon / endothelial cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Breast Invasive Carcinoma (validation): 42/50 (84.0%); Testicular Germ Cell Tumor (TCGA): 101/133 (75.9%)

Cancer IHC. colorectal cancer: 2/9 (22.2%); testis cancer: 2/9 (22.2%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96M89 | HPA portal (live; not the pinned values)

CCDC144A | Unsafe / Protein

ENSG00000170160

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebellum / cells in granular layer: Low (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); nasopharynx / respiratory epithelial cells: Low (Enhanced); stomach 2 / glandular cells: Low (Enhanced); urinary bladder / urothelial cells: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 203/486 (41.8%); Rectum Adenocarcinoma (validation): 85/207 (41.1%); Pancreatic Adenocarcinoma (validation): 32/80 (40.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May play a role in preventing the formation of kidney stones through inhibition of calcium oxalate monohydrate (COM) crystallization, attenuating COM-induced apoptotic injury to renal epithelial cells (PubMed:32991878). May exhibit antilithiatic (preventing the formation of kidney stones) activity through crystal binding, hindering the crystal attachment to renal epithelial cells, a pre-requisite to initiate inflammatory response ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A2RUR9 | HPA portal (live; not the pinned values)

CCDC148 | Unsafe / RNA

ENSG00000153237

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.9 nTPM (limit 2); kidney: 3 nTPM (limit 2); retina: 2.5 nTPM (limit 2); choroid plexus: 2.9 nTPM (baseline 2); kidney: 3 nTPM (baseline 2); retina: 2.5 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 39/58 (67.2%); Breast Invasive Carcinoma (validation): 20/50 (40.0%); Pancreatic Adenocarcinoma (validation): 11/80 (13.8%)

Cancer IHC. colorectal cancer: 2/11 (18.2%); breast cancer: 1/10 (10.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NFR7 | HPA portal (live; not the pinned values)

CCDC15 | Unsafe / RNA

ENSG00000149548

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. bone marrow: 2.1 nTPM (limit 2); bone marrow: 2.1 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 24/50 (48.0%); Pancreatic Adenocarcinoma (validation): 7/80 (8.8%)

Cancer IHC. breast cancer: 11/11 (100.0%); endometrial cancer: 11/11 (100.0%)

Biology. Plays an important role in primary cilium assembly, maintenance, and length regulation. Interacts with centriole inner scaffold proteins to promote proper centriole size and integrity and assembly of functional cilia. Required for the recruitment of both the inner scaffold protein POC1B and the distal SFI1/CETN2 complex to centrioles

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q0P6D6 | HPA portal (live; not the pinned values)

CCDC150 | Unsafe / RNA

ENSG00000144395

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 2.5 nTPM (limit 2); kidney: 2.5 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 26/50 (52.0%); Glioblastoma Multiforme (validation): 17/58 (29.3%); Pancreatic Adenocarcinoma (validation): 21/80 (26.2%)

Cancer IHC. colorectal cancer: 11/12 (91.7%); liver cancer: 9/11 (81.8%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCX0 | HPA portal (live; not the pinned values)

CCDC152 | Unsafe / Protein

ENSG00000198865

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 168/176 (95.5%)

Cancer IHC. pancreatic cancer: 6/11 (54.5%); colorectal cancer: 6/12 (50.0%)

Biology. Detected in stomach

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4G0S7 | HPA portal (live; not the pinned values)

CCDC157 | Unsafe / RNA

ENSG00000187860

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 4 nTPM (limit 2); basal ganglia: 4.5 nTPM (limit 2); cerebral cortex: 4.4 nTPM (limit 2); choroid plexus: 6.2 nTPM (limit 2); hippocampal formation: 4.6 nTPM (limit 2); hypothalamus: 6 nTPM (limit 2); kidney: 2.9 nTPM (limit 2); lung: 2.1 nTPM (limit 2); midbrain: 2.5 nTPM (limit 2); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 40/50 (80.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 88/176 (50.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 89/282 (31.6%)

Cancer IHC. skin cancer: 10/11 (90.9%); stomach cancer: 5/6 (83.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q569K6 | HPA portal (live; not the pinned values)

CCDC169 | Unsafe / RNA

ENSG00000242715

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.8 nTPM (limit 2); retina: 2.5 nTPM (limit 2); bone marrow: 2.8 nTPM (baseline 2); retina: 2.5 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 19/58 (32.8%); Colon Adenocarcinoma (validation): 61/486 (12.6%); Rectum Adenocarcinoma (validation): 23/207 (11.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NNP5 | HPA portal (live; not the pinned values)

CCDC170 | Unsafe / Protein

ENSG00000120262

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (ciliary rootlets): High (Enhanced); nasopharynx / ciliated cells (ciliary rootlets): Medium (Enhanced); smooth muscle / smooth muscle cells: Low (Enhanced); thyroid gland / glandular cells: Low (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (TCGA): 542/1022 (53.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 53/176 (30.1%); Pancreatic Adenocarcinoma (validation): 23/80 (28.7%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Plays a role in Golgi-associated microtubules organization and stabilization

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYT3 | HPA portal (live; not the pinned values)

CCDC171 | Unsafe / Protein

ENSG00000164989

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); cerebellum / Purkinje cells: Low (Uncertain); cerebral cortex / neuropil: Low (Uncertain); colon / endothelial cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 39/58 (67.2%); Rectum Adenocarcinoma (validation): 80/207 (38.6%); Colon Adenocarcinoma (validation): 150/486 (30.9%)

Cancer IHC. colorectal cancer: 4/12 (33.3%); breast cancer: 3/11 (27.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6TFL3 | HPA portal (live; not the pinned values)

CCDC172 | Unsafe / RNA

ENSG00000182645

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 3 nTPM (limit 2); retina: 3 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 5/133 (3.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P0C7W6 | HPA portal (live; not the pinned values)

CCDC18 | Unsafe / Protein

ENSG00000122483

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / Purkinje cells: Medium (Uncertain); cerebral cortex / endothelial cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 49/50 (98.0%); Colon Adenocarcinoma (validation): 369/486 (75.9%)

Cancer IHC. colorectal cancer: 9/12 (75.0%); liver cancer: 7/12 (58.3%)

Biology. May promote pre-adipocyte differentiation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T9S5 | HPA portal (live; not the pinned values)

CCDC181 | Unsafe / Protein

ENSG00000117477

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cilia axoneme): High (Enhanced); nasopharynx / ciliated cells (tip of cilia): High (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 90/141 (63.8%); Testicular Germ Cell Tumor (TCGA): 41/133 (30.8%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Microtubule-binding protein that is essential for the integrity and function of the manchette, facilitating proper sperm head shaping and subsequent flagellum formation during spermatogenesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5TID7 | HPA portal (live; not the pinned values)

CCDC188 | Unsafe / RNA

ENSG00000234409

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 2.5 nTPM (limit 2); hypothalamus: 2.9 nTPM (limit 2); pituitary gland: 9.3 nTPM (limit 2); cerebellum: 2.5 nTPM (baseline 2); hypothalamus: 2.9 nTPM (baseline 2); pituitary gland: 9.3 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 8/176 (4.5%); Glioblastoma Multiforme (validation): 2/58 (3.4%); Pancreatic Adenocarcinoma (TCGA): 6/176 (3.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt H7C350 | HPA portal (live; not the pinned values)

CCDC196 | Unsafe / High RNA

ENSG00000196553

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 103.9 nTPM; liver/hepatocytes; hpa_single_cell_tissue; vital; 2.411 TPM; blood/TREG_NAIVE; dice_treg_naive; vital

Cancer RNA. Breast Invasive Carcinoma (TCGA): 8/1022 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0A1B0GTZ2 | HPA portal (live; not the pinned values)

CCDC34 | Unsafe / RNA

ENSG00000109881

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 5.1 nTPM (limit 2); amygdala: 8.2 nTPM (limit 2); basal ganglia: 10.1 nTPM (limit 2); bone marrow: 13.6 nTPM (limit 2); cerebellum: 2.9 nTPM (limit 2); cerebral cortex: 8.9 nTPM (limit 2); choroid plexus: 21.7 nTPM (limit 2); colon: 8.5 nTPM (limit 6); duodenum: 10.4 nTPM (limit 2); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Rectum Adenocarcinoma (TCGA): 88/88 (100.0%)

Cancer IHC. skin cancer: 8/11 (72.7%); urothelial cancer: 8/11 (72.7%)

Biology. Involved in spermatogenesis. Has a probable role in anterograde intraflagellar transport which is essential for the formation of sperm flagella

Curated tissue note. Expressed in sperm

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96HJ3 | HPA portal (live; not the pinned values)

CCDC38 | Unsafe / RNA

ENSG00000165972

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. liver: 2.7 nTPM (limit 2); liver: 2.7 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 8/176 (4.5%); Testicular Germ Cell Tumor (TCGA): 5/133 (3.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 3/282 (1.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Essential for male fertility. Required for sperm flagellum biogenesis. Also required for acrosome biogenesis. Required for the attachment of developing acrosomes to the nucleus during spermiogenesis and may be involved in the transport of fibrous sheath components

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q502W7 | HPA portal (live; not the pinned values)

CCDC63 | Unsafe / RNA

ENSG00000173093

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skeletal muscle: 2.6 nTPM (limit 2); skeletal muscle: 2.6 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays a role in spermiogenesis. Involved in the elongation of flagella and the formation of sperm heads

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NA47 | HPA portal (live; not the pinned values)

CCDC65 | Unsafe / RNA

ENSG00000139537

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.1 nTPM (limit 2); basal ganglia: 3.4 nTPM (limit 2); cerebral cortex: 2.1 nTPM (limit 2); choroid plexus: 18.9 nTPM (limit 2); hippocampal formation: 3 nTPM (limit 2); hypothalamus: 3.4 nTPM (limit 2); lung: 2.8 nTPM (limit 2); pituitary gland: 2.1 nTPM (limit 2); retina: 6.9 nTPM ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 22/50 (44.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 119/282 (42.2%); Glioblastoma Multiforme (validation): 19/58 (32.8%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); pancreatic cancer: 12/12 (100.0%)

Biology. Component of the nexin-dynein regulatory complex (N-DRC), a key regulator of ciliary/flagellar motility which maintains the alignment and integrity of the distal axoneme and regulates microtubule sliding in motile axonemes (By similarity). Plays a critical role in the assembly of N-DRC and also stabilizes the assembly of multiple inner dynein arms and radial spokes. Coassembles with DRC1 to form a central scaffold needed for assembly ... [full text in table]

Curated tissue note. Highly expressed in adult testis, in spermatocytes and spermatids. Also observed in spermatogonia. Not detected in Leydig cells, nor in fetal testis (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IXS2 | HPA portal (live; not the pinned values)

CCDC7 | Unsafe / RNA

ENSG00000216937

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.6 nTPM (limit 2); retina: 2.6 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 52/58 (89.7%); Pancreatic Adenocarcinoma (validation): 68/80 (85.0%); Breast Invasive Carcinoma (validation): 26/50 (52.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May play a role in tumorigenesis

Curated tissue note. Expressed in epithelium of normal cervix and cervical cancer. Overexpressed in early and interim cervical cancer

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96M83 | HPA portal (live; not the pinned values)

CCDC89 | Unsafe / Protein

ENSG00000179071

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); hippocampus / neuronal cells: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); lymph node / non-germinal center cells: Low (Enhanced); nasopharynx / basal cells: Low (Enhanced); nasopharynx / ciliated cells (cell body): Medium (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 38/58 (65.5%); Breast Invasive Carcinoma (validation): 22/50 (44.0%); Kidney Renal Clear Cell Carcinoma (validation): 12/100 (12.0%)

Cancer IHC. glioma: 1/12 (8.3%); prostate cancer: 1/12 (8.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N998 | HPA portal (live; not the pinned values)

CCDC93 | Unsafe / Protein

ENSG00000125633

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: High (Approved); cerebral cortex / endothelial cells: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. liver cancer: 12/12 (100.0%); colorectal cancer: 11/11 (100.0%)

Biology. Component of the commander complex that is essential for endosomal recycling of transmembrane cargos; the commander complex is composed of composed of the CCC subcomplex and the retriever subcomplex (PubMed:37172566, PubMed:38459129). Component of the CCC complex, which is involved in the regulation of endosomal recycling of surface proteins, including integrins, signaling receptor and channels (PubMed:37172566, PubMed:38459129). The ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q567U6 | HPA portal (live; not the pinned values)

CCHCR1 | Unsafe / RNA

ENSG00000204536

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 10.1 nTPM (limit 10); adrenal gland: 7.3 nTPM (limit 2); amygdala: 9.3 nTPM (limit 2); basal ganglia: 10.5 nTPM (limit 2); cerebellum: 16.8 nTPM (limit 2); cerebral cortex: 10.2 nTPM (limit 2); colon: 15.7 nTPM (limit 6); duodenum: 9.6 nTPM (limit 2); esophagus: 14.7 nTPM (limit 6); ... [full text in table]

Cancer RNA. Ovary Serous Cystadenocarcinoma (validation): 81/81 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 79/80 (98.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be a regulator of keratinocyte proliferation or differentiation

Curated tissue note. Found in all tissues tested, abundantly expressed in heart, liver, skeletal muscle, kidney and pancreas, and to a lesser extent in lung and placenta. Overexpressed in keratinocytes of psoriatic lesions

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TD31 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CCL17 | Unsafe / Protein

ENSG00000102970

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in molecular layer: Low (Uncertain); cerebellum / Purkinje cells: ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (TCGA): 101/176 (57.4%); Breast Invasive Carcinoma (validation): 25/50 (50.0%); Lung Adenocarcinoma (TCGA): 231/497 (46.5%)

Cancer IHC. colorectal cancer: 10/11 (90.9%); endometrial cancer: 10/11 (90.9%)

Biology. Chemokine, which displays chemotactic activity for T lymphocytes, preferentially Th2 cells, but not monocytes or granulocytes. Therefore plays an important role in a wide range of inflammatory and immunological processes (PubMed:8702936, PubMed:9169480). Acts by binding to CCR4 at T-cell surface (PubMed:10540332, PubMed:9169480). Mediates GM-CSF/CSF2-driven pain and inflammation (PubMed:27525438). In the brain, required to maintain the ... [full text in table]

Curated tissue note. Constitutively expressed in thymus. Detected at lower levels in the lung, colon and small intestine (PubMed:8702936). Expressed in stimulated peripheral blood mononuclear cells, but not in resting cells ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92583 | HPA portal (live; not the pinned values)

CCL27 | Unsafe / Protein

ENSG00000213927

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); lymph node / non-germinal center cells: Medium (Enhanced); skin 1 / cells in spinous layer: Medium (Enhanced); skin 2 / cells in spinous layer: Medium (Enhanced); skin 2 / fibrohistiocytic cells: High (Enhanced); spleen / cells in red pulp: ... [full text in table]

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 35/492 (7.1%); Bladder Urothelial Carcinoma (TCGA): 11/169 (6.5%); Skin Cuteneous Melanoma (TCGA): 6/99 (6.1%)

Cancer IHC. renal cancer: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. Chemotactic factor that attracts skin-associated memory T-lymphocytes. May play a role in mediating homing of lymphocytes to cutaneous sites. Binds to CCR10

Curated tissue note. Testis, thymus, placenta, ovary and skin

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y4X3 | HPA portal (live; not the pinned values)

CCN6 | Unsafe / RNA

ENSG00000112761

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 3.1 nTPM (limit 2); cerebellum: 3.1 nTPM (baseline 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (validation): 33/81 (40.7%); Pancreatic Adenocarcinoma (validation): 25/80 (31.2%); Ovary Serous Cystadenocarcinoma (TCGA): 106/349 (30.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays a role in mitochondrial electron transport and mitochondrial respiration (PubMed:27252383). Through its regulation of the mitochondrial function may play a role in normal postnatal skeletal growth and cartilage homeostasis (PubMed:10471507, PubMed:27252383)

Curated tissue note. Predominant expression in adult kidney and testis and fetal kidney. Weaker expression found in placenta, ovary, prostate and small intestine (PubMed:10471507, PubMed:9843955). Also expressed in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95389 | HPA portal (live; not the pinned values)

CCNA1 | Unsafe / Protein

ENSG00000133101

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): High (Enhanced); nasopharynx / ciliated cells (cell body): Medium (Enhanced)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 265/349 (75.9%); Ovary Serous Cystadenocarcinoma (validation): 58/81 (71.6%); Uterine Corpus Endometrial Carcinoma (TCGA): 111/176 (63.1%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May be involved in the control of the cell cycle at the G1/S (start) and G2/M (mitosis) transitions. May primarily function in the control of the germline meiotic cell cycle and additionally in the control of mitotic cell cycle in some somatic cells

Curated tissue note. Very high levels in testis and very low levels in brain. Also found in myeloid leukemia cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P78396 | HPA portal (live; not the pinned values)

CCNB2 | Unsafe / Protein

ENSG00000157456

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: High (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); colon / endocrine cells: High (Enhanced); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (TCGA): 88/88 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 491/492 (99.8%)

Cancer IHC. colorectal cancer: 10/12 (83.3%); cervical cancer: 8/12 (66.7%)

Biology. Essential for the control of the cell cycle at the G2/M (mitosis) transition

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95067 | HPA portal (live; not the pinned values)

CCNB3 | Unsafe / RNA

ENSG00000147082

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. spinal cord: 2.5 nTPM (limit 2); spinal cord: 2.5 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 11/50 (22.0%); Glioblastoma Multiforme (validation): 2/58 (3.4%); Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Cyclins are positive regulatory subunits of the cyclin-dependent kinases (CDKs), and thereby play an essential role in the control of the cell cycle, notably via their destruction during cell division. Its tissue specificity suggest that it may be required during early meiotic prophase I

Curated tissue note. Testis specific. In testis, it is expressed in developing germ cells, but not in Leydig cells. Weakly or not expressed in other tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WWL7 | HPA portal (live; not the pinned values)

CCNE2 | Unsafe / Protein

ENSG00000175305

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); caudate / glial cells: Medium (Enhanced); cerebellum / cells in granular layer: Low (Enhanced); cerebellum / cells in molecular layer: Medium (Enhanced); cerebral cortex / glial cells: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 480/486 (98.8%); Rectum Adenocarcinoma (validation): 199/207 (96.1%)

Cancer IHC. glioma: 7/12 (58.3%); lymphoma: 7/12 (58.3%)

Biology. Essential for the control of the cell cycle at the late G1 and early S phase

Curated tissue note. According to PubMed:9858585, highest levels of expression in adult testis, thymus and brain. Lower levels in placenta, spleen and colon. Consistently elevated levels in tumor-derived cells compared to ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O96020 | HPA portal (live; not the pinned values)

CCSER1 | Unsafe / Protein

ENSG00000184305

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); cerebral cortex / endothelial cells: Low (Uncertain); colon / endothelial cells: Medium (Uncertain); duodenum / glandular cells: Low (Uncertain); lung / alveolar cells: Medium (Uncertain); lung / macrophages: Medium (Uncertain); rectum / glandular ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 72/80 (90.0%); Colon Adenocarcinoma (validation): 55/486 (11.3%); Glioblastoma Multiforme (validation): 6/58 (10.3%)

Cancer IHC. colorectal cancer: 11/12 (91.7%); prostate cancer: 8/11 (72.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9C0I3 | HPA portal (live; not the pinned values)

CCT6B | Unsafe / Protein

ENSG00000132141

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 47/50 (94.0%); Glioblastoma Multiforme (validation): 40/58 (69.0%); Prostate Adenocarcinoma (TCGA): 185/480 (38.5%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); testis cancer: 11/12 (91.7%)

Biology. Could function as a component of a tissue-specific chaperonin-containing T-complex (TRiC), a molecular chaperone complex that assists the folding of actin, tubulin and other proteins upon ATP hydrolysis

Curated tissue note. Testis-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92526 | HPA portal (live; not the pinned values)

CD101 | Unsafe / Protein

ENSG00000134256

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 47/58 (81.0%); Breast Invasive Carcinoma (validation): 23/50 (46.0%); Rectum Adenocarcinoma (validation): 35/207 (16.9%)

Cancer IHC. melanoma: 7/12 (58.3%); urothelial cancer: 5/12 (41.7%)

Biology. Plays a role as inhibitor of T-cells proliferation induced by CD3. Inhibits expression of IL2RA on activated T-cells and secretion of IL2. Inhibits tyrosine kinases that are required for IL2 production and cellular proliferation. Inhibits phospholipase C-gamma-1/PLCG1 phosphorylation and subsequent CD3-induced changes in intracellular free calcium. Prevents nuclear translocation of nuclear factor of activated T-cell to the nucleus. ... [full text in table]

Curated tissue note. Expressed in lung, thymus and small intestine. Detected in cutaneous dendritic cells, activated T-cells, monocytes and granulocytes as well as with epithelial cells with dendritic morphology. Expressed in some ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q93033 | HPA portal (live; not the pinned values)

CD200R1L | Unsafe / High RNA

ENSG00000206531

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 110.7 nTPM; blood/basophil; hpa_immune_cell_samples; vital; 167.4 TPM; blood/basophil; hpa_immune_monaco; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%); Colon Adenocarcinoma (validation): 4/486 (0.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be a receptor for the CD200/OX2 cell surface glycoprotein

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6Q8B3 | HPA portal (live; not the pinned values)

CDADC1 | Unsafe / Protein

ENSG00000102543

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Uncertain); lymph node / germinal center cells: Medium (Uncertain); lymph node / non-germinal center cells: Medium (Uncertain); spleen / cells in red pulp: Medium (Uncertain); spleen / cells in white pulp: Medium (Uncertain)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Pancreatic Adenocarcinoma (validation): 51/80 (63.7%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Catalyzes the deamination of dCTP to dUTP, exhibiting a strong substrate preference for dCTP over CTP (PubMed:40324085, PubMed:40504152). No deaminase activity toward 5-methyl-dCTP, cytidine, or deoxycytidine (PubMed:40324085, PubMed:40504152). May play an important role in testicular development and spermatogenesis (PubMed:16955368). In addition to its role on endogenous nucleotides, it deaminates the active triphosphate forms of the ... [full text in table]

Curated tissue note. Widely expressed. Expressed at high levels in the testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BWV3 | HPA portal (live; not the pinned values)

CDC25A | Unsafe / RNA

ENSG00000164045

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 15.1 nTPM (limit 2); bone marrow: 15.1 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 127/133 (95.5%); Colon Adenocarcinoma (validation): 462/486 (95.1%); Glioblastoma Multiforme (validation): 54/58 (93.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Tyrosine protein phosphatase which functions as a dosage-dependent inducer of mitotic progression (PubMed:12676925, PubMed:14559997, PubMed:1836978, PubMed:20360007). Directly dephosphorylates CDK1 and stimulates its kinase activity (PubMed:20360007). Also dephosphorylates CDK2 in complex with cyclin-E, in vitro (PubMed:20360007)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P30304 | HPA portal (live; not the pinned values)

CDC25C | Unsafe / Protein

ENSG00000158402

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 449/486 (92.4%); Rectum Adenocarcinoma (validation): 177/207 (85.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 236/283 (83.4%)

Cancer IHC. cervical cancer: 12/12 (100.0%); lung cancer: 12/12 (100.0%)

Biology. Functions as a dosage-dependent inducer in mitotic control. Tyrosine protein phosphatase required for progression of the cell cycle (PubMed:8119945). When phosphorylated, highly effective in activating G2 cells into prophase (PubMed:8119945). Directly dephosphorylates CDK1 and activates its kinase activity (PubMed:8119945)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P30307 | HPA portal (live; not the pinned values)

CDC40 | Unsafe / RNA

ENSG00000168438

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 10.7 nTPM (limit 10); adrenal gland: 14.2 nTPM (limit 2); amygdala: 7.5 nTPM (limit 2); basal ganglia: 14.2 nTPM (limit 2); bone marrow: 37.7 nTPM (limit 2); cerebellum: 8.6 nTPM (limit 2); cerebral cortex: 12.9 nTPM (limit 2); choroid plexus: 6.6 nTPM (limit 2); colon: 10.7 nTPM ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Required for pre-mRNA splicing as component of the activated spliceosome (PubMed:33220177). Plays an important role in embryonic brain development; this function does not require proline isomerization (PubMed:33220177)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O60508 | HPA portal (live; not the pinned values)

CDC45 | Unsafe / Protein

ENSG00000093009

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); colon / endothelial cells: Low (Enhanced); hippocampus / glial cells: Low (Enhanced); kidney / cells in glomeruli: Low (Enhanced); kidney / cells ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 66/68 (97.1%); Lung Squamous Cell Carcinoma (TCGA): 474/489 (96.9%); Colon Adenocarcinoma (TCGA): 244/254 (96.1%)

Cancer IHC. prostate cancer: 6/11 (54.5%); melanoma: 4/11 (36.4%)

Biology. Required for initiation of chromosomal DNA replication. Core component of CDC45-MCM-GINS (CMG) helicase, the molecular machine that unwinds template DNA during replication, and around which the replisome is built

Curated tissue note. Widely expressed, highest levels are found in adult testis and thymus and in fetal liver

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75419 | HPA portal (live; not the pinned values)

CDC6 | Unsafe / Protein

ENSG00000094804

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%); Colon Adenocarcinoma (TCGA): 253/254 (99.6%)

Cancer IHC. testis cancer: 12/12 (100.0%); pancreatic cancer: 9/11 (81.8%)

Biology. Involved in the initiation of DNA replication. Also participates in checkpoint controls that ensure DNA replication is completed before mitosis is initiated

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q99741 | HPA portal (live; not the pinned values)

CDCA2 | Unsafe / Protein

ENSG00000184661

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); colon / endocrine cells: High (Enhanced); colon / enterocytes: High (Enhanced); colon / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Colon Adenocarcinoma (validation): 434/486 (89.3%); Rectum Adenocarcinoma (validation): 170/207 (82.1%)

Cancer IHC. colorectal cancer: 4/11 (36.4%); lung cancer: 3/11 (27.3%)

Biology. Regulator of chromosome structure during mitosis required for condensin-depleted chromosomes to retain their compact architecture through anaphase. Acts by mediating the recruitment of phosphatase PP1-gamma subunit (PPP1CC) to chromatin at anaphase and into the following interphase. At anaphase onset, its association with chromatin targets a pool of PPP1CC to dephosphorylate substrates

Curated tissue note. Ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q69YH5 | HPA portal (live; not the pinned values)

CDCA5 | Unsafe / Protein

ENSG00000146670

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: High (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); cerebellum / Purkinje cells: High (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); colon / endocrine cells: High ... [full text in table]

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 283/283 (100.0%); Rectum Adenocarcinoma (TCGA): 88/88 (100.0%); Colon Adenocarcinoma (TCGA): 253/254 (99.6%)

Cancer IHC. liver cancer: 12/12 (100.0%); lung cancer: 12/12 (100.0%)

Biology. Regulator of sister chromatid cohesion in mitosis, stabilizing cohesin complex association with chromatin. May antagonize the action of WAPL which stimulates cohesin dissociation from chromatin. Cohesion ensures that chromosome partitioning is accurate in both meiotic and mitotic cells and plays an important role in DNA repair. Required for efficient DNA double-stranded break repair

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96FF9 | HPA portal (live; not the pinned values)

CDCA8 | Unsafe / Protein

ENSG00000134690

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); cerebellum / Purkinje cells: Low (Enhanced); cerebral cortex / neuropil: Low (Enhanced); colon / ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (TCGA): 88/88 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); cervical cancer: 11/12 (91.7%)

Biology. Component of the chromosomal passenger complex (CPC), a complex that acts as a key regulator of mitosis. The CPC complex has essential functions at the centromere in ensuring correct chromosome alignment and segregation and is required for chromatin-induced microtubule stabilization and spindle assembly. Major effector of the TTK kinase in the control of attachment-error-correction and chromosome alignment

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q53HL2 | HPA portal (live; not the pinned values)

CDK13 | Unsafe / Protein

ENSG00000065883

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Uncertain); duodenum / glandular cells: High (Uncertain); esophagus / squamous epithelial cells: Low (Uncertain); heart muscle / cardiomyocytes: Low (Uncertain); kidney / cells in tubules: High (Uncertain); oral mucosa / squamous epithelial cells: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. cervical cancer: 2/10 (20.0%); colorectal cancer: 2/10 (20.0%)

Biology. Cyclin-dependent kinase which displays CTD kinase activity and is required for RNA splicing. Has CTD kinase activity by hyperphosphorylating the C-terminal heptapeptide repeat domain (CTD) of the largest RNA polymerase II subunit RPB1, thereby acting as a key regulator of transcription elongation. Required for RNA splicing, probably by phosphorylating SRSF1/SF2. Required during hematopoiesis. In case of infection by HIV-1 virus, ... [full text in table]

Curated tissue note. Expressed in fetal brain, liver, muscle and in adult brain. Also expressed in neuroblastoma and glioblastoma tumors

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14004 | HPA portal (live; not the pinned values)

CDKL3 | Unsafe / RNA

ENSG00000006837

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.8 nTPM (limit 2); cerebral cortex: 2.4 nTPM (limit 2); heart muscle: 2.4 nTPM (limit 2); hippocampal formation: 2.6 nTPM (limit 2); hypothalamus: 2.7 nTPM (limit 2); midbrain: 3 nTPM (limit 2); retina: 4.2 nTPM (limit 2); skeletal muscle: 3.1 nTPM (limit 2); spinal cord: 2.9 nTPM (limit ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 42/50 (84.0%); Glioblastoma Multiforme (validation): 35/58 (60.3%); Pancreatic Adenocarcinoma (validation): 36/80 (45.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IVW4 | HPA portal (live; not the pinned values)

CDKN3 | Unsafe / RNA

ENSG00000100526

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.1 nTPM (limit 2); basal ganglia: 2.7 nTPM (limit 2); bone marrow: 42.3 nTPM (limit 2); cerebellum: 2.9 nTPM (limit 2); cerebral cortex: 4.4 nTPM (limit 2); choroid plexus: 12 nTPM (limit 2); colon: 13.5 nTPM (limit 6); duodenum: 12.2 nTPM (limit 2); esophagus: 9.9 nTPM (limit 6); heart ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (TCGA): 88/88 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May play a role in cell cycle regulation. Dual specificity CC phosphatase active toward substrates containing either phosphotyrosine or phosphoserine residues (PubMed:8127873, PubMed:8242750). Dephosphorylates CDK2 at 'Thr-160' in a cyclin-dependent manner (PubMed:7569954)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q16667 | HPA portal (live; not the pinned values)

CDRT15L2 | Unsafe / RNA

ENSG00000214819

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. hippocampal formation: 2.2 nTPM (limit 2); hippocampal formation: 2.2 nTPM (baseline 2)

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 10/283 (3.5%); Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 3/176 (1.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A8MXV6 | HPA portal (live; not the pinned values)

CDRT4 | Unsafe / Protein

ENSG00000239704

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / Purkinje cells: Medium (Uncertain); colon / endothelial cells: Low (Uncertain); colon / glandular cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 44/50 (88.0%); Colon Adenocarcinoma (validation): 335/486 (68.9%); Rectum Adenocarcinoma (validation): 142/207 (68.6%)

Cancer IHC. liver cancer: 5/12 (41.7%); pancreatic cancer: 3/10 (30.0%)

Biology. Expressed in fetal skeletal muscle and kidney

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N9R6 | HPA portal (live; not the pinned values)

CELA3B | Unsafe / Protein

ENSG00000219073

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. pancreas / exocrine glandular cells: High (Supported)

Cancer RNA. Pancreatic Adenocarcinoma (TCGA): 97/176 (55.1%); Pancreatic Adenocarcinoma (validation): 24/80 (30.0%); Testicular Germ Cell Tumor (TCGA): 5/133 (3.8%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Efficient protease with alanine specificity but only little elastolytic activity

Curated tissue note. Pancreas. Not detectable in keratinocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P08861 | HPA portal (live; not the pinned values)

CENPA | Unsafe / Protein

ENSG00000115163

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); colon / endothelial cells: Medium (Enhanced); colon / glandular cells: Medium (Enhanced); duodenum / glandular cells: Medium (Enhanced); kidney / cells in glomeruli: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); lymph node / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 52/58 (89.7%); Colon Adenocarcinoma (TCGA): 224/254 (88.2%); Ovary Serous Cystadenocarcinoma (TCGA): 307/349 (88.0%)

Cancer IHC. cervical cancer: 6/12 (50.0%); colorectal cancer: 5/12 (41.7%)

Biology. Histone H3-like nucleosomal protein that is specifically found in centromeric nucleosomes (PubMed:11756469, PubMed:14667408, PubMed:15282608, PubMed:15475964, PubMed:15702419, PubMed:17651496, PubMed:19114591, PubMed:20739937, PubMed:27499292, PubMed:7962047, PubMed:9024683). Replaces conventional H3 in the nucleosome core of centromeric chromatin that serves as an assembly site for the inner kinetochore (PubMed:18072184). The presence ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P49450 | HPA portal (live; not the pinned values)

CENPE | Unsafe / Protein

ENSG00000138778

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: High (Enhanced); duodenum / glandular cells: Low (Enhanced); esophagus / squamous epithelial cells: Medium (Enhanced); heart muscle / cardiomyocytes: Medium (Enhanced); kidney / distal tubules: High ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Colon Adenocarcinoma (validation): 455/486 (93.6%); Breast Invasive Carcinoma (validation): 43/50 (86.0%)

Cancer IHC. skin cancer: 9/12 (75.0%); testis cancer: 7/11 (63.6%)

Biology. Microtubule plus-end-directed kinetochore motor which plays an important role in chromosome congression, microtubule-kinetochore conjugation and spindle assembly checkpoint activation. Drives chromosome congression (alignment of chromosomes at the spindle equator resulting in the formation of the metaphase plate) by mediating the lateral sliding of polar chromosomes along spindle microtubules towards the spindle equator and by aiding ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q02224 | HPA portal (live; not the pinned values)

CENPH | Unsafe / Protein

ENSG00000153044

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / Purkinje cells: High (Approved); cerebral cortex / neuronal cells: Low (Approved); cerebral cortex / neuropil: Low (Approved); colon / glandular cells: Low (Approved); duodenum / glandular ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 253/254 (99.6%); Rectum Adenocarcinoma (TCGA): 87/88 (98.9%); Colon Adenocarcinoma (validation): 480/486 (98.8%)

Cancer IHC. colorectal cancer: 4/9 (44.4%); stomach cancer: 3/11 (27.3%)

Biology. Component of the CENPA-NAC (nucleosome-associated) complex, a complex that plays a central role in assembly of kinetochore proteins, mitotic progression and chromosome segregation. The CENPA-NAC complex recruits the CENPA-CAD (nucleosome distal) complex and may be involved in incorporation of newly synthesized CENPA into centromeres. Required for chromosome congression and efficiently align the chromosomes on a metaphase plate

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H3R5 | HPA portal (live; not the pinned values)

CENPI | Unsafe / RNA

ENSG00000102384

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 5.5 nTPM (limit 2); duodenum: 2.4 nTPM (limit 2); bone marrow: 5.5 nTPM (baseline 2); duodenum: 2.4 nTPM (baseline 2)

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 205/207 (99.0%); Lung Squamous Cell Carcinoma (validation): 66/68 (97.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of the CENPA-CAD (nucleosome distal) complex, a complex recruited to centromeres which is involved in assembly of kinetochore proteins, mitotic progression and chromosome segregation. May be involved in incorporation of newly synthesized CENPA into centromeres via its interaction with the CENPA-NAC complex. Required for the localization of CENPF, MAD1L1 and MAD2 (MAD2L1 or MAD2L2) to kinetochores. Involved in the response of ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92674 | HPA portal (live; not the pinned values)

CENPK | Unsafe / RNA

ENSG00000123219

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 24.6 nTPM (limit 2); colon: 8.7 nTPM (limit 6); duodenum: 8.1 nTPM (limit 2); lymph node: 14.4 nTPM (limit 6); rectum: 11.5 nTPM (limit 6); small intestine: 8.4 nTPM (limit 2); smooth muscle: 3.3 nTPM (limit 2); thymus: 21.7 nTPM (limit 10); bone marrow: 24.6 nTPM (baseline 2); colon: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 484/486 (99.6%); Rectum Adenocarcinoma (validation): 206/207 (99.5%); Lung Squamous Cell Carcinoma (validation): 67/68 (98.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of the CENPA-CAD (nucleosome distal) complex, a complex recruited to centromeres which is involved in assembly of kinetochore proteins, mitotic progression and chromosome segregation. May be involved in incorporation of newly synthesized CENPA into centromeres via its interaction with the CENPA-NAC complex. Acts in coordination with KNL1 to recruit the NDC80 complex to the outer kinetochore

Curated tissue note. Detected in several fetal organs with highest levels in fetal liver. In adults, it is weakly expressed in lung and placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BS16 | HPA portal (live; not the pinned values)

CENPL | Unsafe / Protein

ENSG00000120334

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / Purkinje cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Squamous Cell Carcinoma (validation): 63/68 (92.6%); Colon Adenocarcinoma (validation): 435/486 (89.5%)

Cancer IHC. breast cancer: 6/12 (50.0%); ovarian cancer: 4/10 (40.0%)

Biology. Component of the CENPA-CAD (nucleosome distal) complex, a complex recruited to centromeres which is involved in assembly of kinetochore proteins, mitotic progression and chromosome segregation. May be involved in incorporation of newly synthesized CENPA into centromeres via its interaction with the CENPA-NAC complex

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N0S6 | HPA portal (live; not the pinned values)

CENPO | Unsafe / RNA

ENSG00000138092

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.9 nTPM (limit 2); amygdala: 4.2 nTPM (limit 2); basal ganglia: 4.2 nTPM (limit 2); bone marrow: 9.1 nTPM (limit 2); cerebellum: 4 nTPM (limit 2); cerebral cortex: 4.8 nTPM (limit 2); choroid plexus: 2.3 nTPM (limit 2); duodenum: 3.1 nTPM (limit 2); heart muscle: 2.7 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 482/486 (99.2%); Rectum Adenocarcinoma (validation): 205/207 (99.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of the CENPA-CAD (nucleosome distal) complex, a complex recruited to centromeres which is involved in assembly of kinetochore proteins, mitotic progression and chromosome segregation. May be involved in incorporation of newly synthesized CENPA into centromeres via its interaction with the CENPA-NAC complex. Modulates the kinetochore-bound levels of NDC80 complex

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BU64 | HPA portal (live; not the pinned values)

CENPP | Unsafe / Protein

ENSG00000188312

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Medium (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 191/207 (92.3%); Colon Adenocarcinoma (validation): 447/486 (92.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 443/492 (90.0%)

Cancer IHC. pancreatic cancer: 8/10 (80.0%); testis cancer: 8/10 (80.0%)

Biology. Component of the CENPA-CAD (nucleosome distal) complex, a complex recruited to centromeres which is involved in assembly of kinetochore proteins, mitotic progression and chromosome segregation. May be involved in incorporation of newly synthesized CENPA into centromeres via its interaction with the CENPA-NAC complex

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6IPU0 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CENPS-CORT | Unsafe / RNA

ENSG00000251503

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 4 nTPM (limit 2); basal ganglia: 4.5 nTPM (limit 2); bone marrow: 8.3 nTPM (limit 2); cerebral cortex: 3.8 nTPM (limit 2); hippocampal formation: 3.2 nTPM (limit 2); hypothalamus: 3.5 nTPM (limit 2); kidney: 2.2 nTPM (limit 2); midbrain: 2.9 nTPM (limit 2); pituitary gland: 2.4 nTPM ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 91/133 (68.4%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 155/283 (54.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 68/176 (38.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. DNA-binding component of the Fanconi anemia (FA) core complex. Required for the normal activation of the FA pathway, leading to monoubiquitination of the FANCI-FANCD2 complex in response to DNA damage, cellular resistance to DNA cross-linking drugs, and prevention of chromosomal breakage (PubMed:20347428, PubMed:20347429). In complex with CENPX (MHF heterodimer), crucial cofactor for FANCM in both binding and ATP-dependent remodeling ... [full text in table]

Curated tissue note. Ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N2Z9 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CENPU | Unsafe / RNA

ENSG00000151725

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 61.8 nTPM (limit 2); colon: 7.5 nTPM (limit 6); duodenum: 4.9 nTPM (limit 2); esophagus: 10.1 nTPM (limit 6); liver: 3.6 nTPM (limit 2); lymph node: 15.8 nTPM (limit 6); rectum: 7.8 nTPM (limit 6); skin: 14.9 nTPM (limit 10); small intestine: 5.2 nTPM (limit 2); smooth muscle: 2.6 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Lung Squamous Cell Carcinoma (validation): 66/68 (97.1%); Colon Adenocarcinoma (validation): 464/486 (95.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of the CENPA-NAC (nucleosome-associated) complex, a complex that plays a central role in assembly of kinetochore proteins, mitotic progression and chromosome segregation. The CENPA-NAC complex recruits the CENPA-CAD (nucleosome distal) complex and may be involved in incorporation of newly synthesized CENPA into centromeres. Plays an important role in the correct PLK1 localization to the mitotic kinetochores. A scaffold ... [full text in table]

Curated tissue note. Expressed at high levels in the testis, fetal liver, thymus, bone marrow and at lower levels in the lymph nodes, placenta, colon and spleen. Present in all cell lines examined, including B-cells, T-cells, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q71F23 | HPA portal (live; not the pinned values)

CENPVL1 | Unsafe / RNA

ENSG00000223591

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 3.9 nTPM (limit 2); hypothalamus: 3.9 nTPM (limit 2); basal ganglia: 3.9 nTPM (baseline 2); hypothalamus: 3.9 nTPM (baseline 2)

Cancer RNA. Liver Hepatocellular Carcinoma (TCGA): 3/362 (0.8%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0A0U1RR11 | UniProt P0DPI3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CENPVL2 | Unsafe / RNA

ENSG00000283093

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 4.7 nTPM (limit 2); hypothalamus: 4.6 nTPM (limit 2); basal ganglia: 4.7 nTPM (baseline 2); hypothalamus: 4.6 nTPM (baseline 2)

Cancer RNA. Liver Hepatocellular Carcinoma (TCGA): 3/362 (0.8%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0A0U1RR11 | UniProt P0DPI3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CEP120 | Unsafe / RNA

ENSG00000168944

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 6.8 nTPM (limit 2); amygdala: 4.5 nTPM (limit 2); basal ganglia: 4.9 nTPM (limit 2); bone marrow: 2.9 nTPM (limit 2); cerebellum: 11.2 nTPM (limit 2); cerebral cortex: 4.2 nTPM (limit 2); colon: 6.9 nTPM (limit 6); duodenum: 4.7 nTPM (limit 2); heart muscle: 6 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 79/80 (98.8%); Lung Adenocarcinoma (validation): 102/105 (97.1%)

Cancer IHC. cervical cancer: 2/12 (16.7%); endometrial cancer: 2/12 (16.7%)

Biology. Plays a role in the microtubule-dependent coupling of the nucleus and the centrosome. Involved in the processes that regulate centrosome-mediated interkinetic nuclear migration (INM) of neural progenitors and for proper positioning of neurons during brain development. Also implicated in the migration and selfrenewal of neural progenitors. Required for centriole duplication and maturation during mitosis and subsequent ciliogenesis (By ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N960 | HPA portal (live; not the pinned values)

CEP128 | Unsafe / Protein

ENSG00000100629

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 54/58 (93.1%); Colon Adenocarcinoma (validation): 432/486 (88.9%); Rectum Adenocarcinoma (validation): 181/207 (87.4%)

Cancer IHC. liver cancer: 12/12 (100.0%); endometrial cancer: 11/12 (91.7%)

Biology. Is involved in the negative regulation of ciliogenesis and plays a role in spermatogenesis (PubMed:29514088, PubMed:35296684). Contributes to the regulation of the TGFB/BMP signaling pathway at the primary cilium, where it is required for ciliary localization of RAB11A, likely affecting the trafficking and regulation of TGFB/BMP pathway components (PubMed:29514088). Required, together with ODF2, for the formation of the subdistal ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZU80 | HPA portal (live; not the pinned values)

CEP152 | Unsafe / Protein

ENSG00000103995

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Medium (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / glial cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Colon Adenocarcinoma (validation): 457/486 (94.0%); Rectum Adenocarcinoma (validation): 190/207 (91.8%)

Cancer IHC. ovarian cancer: 4/11 (36.4%); pancreatic cancer: 3/11 (27.3%)

Biology. Necessary for centrosome duplication; the function also seems to involve CEP63, CDK5RAP2 and WDR62 through a stepwise assembled complex at the centrosome that recruits CDK2 required for centriole duplication (PubMed:26297806). Acts as a molecular scaffold facilitating the interaction of PLK4 and CPAP, 2 molecules involved in centriole formation (PubMed:20852615, PubMed:21059844). Proposed to snatch PLK4 away from PLK4:CEP92 complexes ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O94986 | HPA portal (live; not the pinned values)

CEP170 | Unsafe / Protein

ENSG00000143702

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebral cortex / neuropil: Low (Uncertain); colon / glandular cells: Low (Uncertain); duodenum / glandular cells: Low (Uncertain); hippocampus / neuronal cells: Low (Uncertain); lung / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. colorectal cancer: 7/11 (63.6%); testis cancer: 4/10 (40.0%)

Biology. Plays a role in microtubule organization (PubMed:15616186). Required for centriole subdistal appendage assembly (PubMed:28422092)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5SW79 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CEP19 | Unsafe / Protein

ENSG00000174007

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced); caudate / glial cells: Medium (Enhanced); cerebellum / cells in molecular layer: Medium (Enhanced); cerebral cortex / glial cells: Medium (Enhanced); cerebral cortex / neuropil: Low (Enhanced); colon / glandular cells: Low (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 88/141 (62.4%); Testicular Germ Cell Tumor (TCGA): 73/133 (54.9%)

Cancer IHC. endometrial cancer: 3/10 (30.0%); testis cancer: 2/9 (22.2%)

Biology. Required for ciliation (PubMed:28428259, PubMed:28625565, PubMed:28659385). Recruits the RABL2B GTPase to the ciliary base to initiate ciliation. After specifically capturing the activated GTP-bound RABL2B, the CEP19-RABL2B complex binds intraflagellar transport (IFT) complex B from the large pool pre-docked at the base of the cilium and thus triggers its entry into the cilia (PubMed:28428259, PubMed:28625565). Involved in the early ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96LK0 | HPA portal (live; not the pinned values)

CEP57L1 | Unsafe / Protein

ENSG00000183137

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. endometrial cancer: 12/12 (100.0%); glioma: 12/12 (100.0%)

Biology. Centrosomal protein which may be required for microtubule attachment to centrosomes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYX8 | HPA portal (live; not the pinned values)

CEP78 | Unsafe / Protein

ENSG00000148019

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / Purkinje cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 48/50 (96.0%); Pancreatic Adenocarcinoma (validation): 75/80 (93.8%)

Cancer IHC. cervical cancer: 8/11 (72.7%); urothelial cancer: 8/12 (66.7%)

Biology. Centriole wall protein that localizes to mature centrioles and regulates centriole and cilia biogenesis (PubMed:27246242, PubMed:27588451, PubMed:28242748, PubMed:34259627). Involved in centrosome duplication: required for efficient PLK4 centrosomal localization and PLK4-induced overduplication of centrioles (PubMed:27246242). Involved in cilium biogenesis and controls cilium length (PubMed:27588451). Acts as a regulator of protein ... [full text in table]

Curated tissue note. Widely expressed (PubMed:27588451, PubMed:27588452). Expressed in different retinal cell types with higher expression in cone compared to rod cells (at protein level) (PubMed:27588452)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JTW2 | HPA portal (live; not the pinned values)

CERS1 | Unsafe / Protein

ENSG00000223802

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); colon / endocrine cells: Medium (Enhanced); colon / enterocytes: Low (Enhanced); heart muscle / cardiomyocytes: Low (Enhanced); hippocampus / neuronal cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Skin Cuteneous Melanoma (TCGA): 96/99 (97.0%); Testicular Germ Cell Tumor (TCGA): 124/133 (93.2%)

Cancer IHC. glioma: 2/10 (20.0%); colorectal cancer: 2/12 (16.7%)

Biology. Ceramide synthase that catalyzes the transfer of the acyl chain from acyl-CoA to a sphingoid base, with high selectivity toward stearoyl-CoA (octadecanoyl-CoA; C18:0-CoA) (PubMed:17977534, PubMed:23530041, PubMed:26887952, PubMed:31916624). N-acylates sphinganine and sphingosine bases to form dihydroceramides and ceramides in de novo synthesis and salvage pathways, respectively (PubMed:17977534, PubMed:23530041, PubMed:24782409, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P27544 | HPA portal (live; not the pinned values)

CERS3 | Unsafe / Protein

ENSG00000154227

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / endothelial cells: ... [full text in table]

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 459/492 (93.3%); Lung Squamous Cell Carcinoma (validation): 50/68 (73.5%); Lung Squamous Cell Carcinoma (TCGA): 331/489 (67.7%)

Cancer IHC. cervical cancer: 7/12 (58.3%); colorectal cancer: 5/12 (41.7%)

Biology. Ceramide synthase that catalyzes the transfer of the acyl chain from acyl-CoA to a sphingoid base, with high selectivity toward very- and ultra-long-chain fatty acyl-CoA (chain length greater than C22) (PubMed:17977534, PubMed:22038835, PubMed:26887952). N-acylates sphinganine and sphingosine bases to form dihydroceramides and ceramides in de novo synthesis and salvage pathways, respectively (PubMed:17977534, PubMed:22038835, ... [full text in table]

Curated tissue note. Expressed in the epidermis, where it localizes at the interface between the stratum granulosum and the stratum corneum (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IU89 | HPA portal (live; not the pinned values)

CFAP100 | Unsafe / Protein

ENSG00000163885

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (tip of cilia): Medium (Enhanced); nasopharynx / ciliated cells (tip of cilia): Medium (Enhanced)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 33/176 (18.8%); Ovary Serous Cystadenocarcinoma (validation): 12/81 (14.8%); Glioblastoma Multiforme (validation): 6/58 (10.3%)

Cancer IHC. testis cancer: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. Microtubule-associated protein that promotes microtubule polymerization and regulates microtubule organization (PubMed:37192300). Plays a critical role in ensuring proper epidermal development and function by regulating mitosis in keratinocytes (PubMed:40593606). Maintains spindle pole integrity during keratinocyte division through the recruitment of the microtubule glutamylase TTLL13 to spindle poles, thereby enhancing localized ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q494V2 | HPA portal (live; not the pinned values)

CFAP107 | Unsafe / RNA

ENSG00000157330

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.1 nTPM (limit 2); choroid plexus: 45.3 nTPM (limit 2); hypothalamus: 2.6 nTPM (limit 2); basal ganglia: 2.1 nTPM (baseline 2); choroid plexus: 45.3 nTPM (baseline 2); hypothalamus: 2.6 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 43/176 (24.4%); Glioblastoma Multiforme (TCGA): 27/141 (19.1%); Glioblastoma Multiforme (validation): 10/58 (17.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia axoneme, which is required for motile cilia beating

Curated tissue note. Expressed in airway epithelial cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N1D5 | HPA portal (live; not the pinned values)

CFAP126 | Unsafe / Protein

ENSG00000188931

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (ciliary rootlets): Low (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cilia axoneme): High (Enhanced); nasopharynx / ciliated cells (ciliary rootlets): Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%); Lung Adenocarcinoma (validation): 104/105 (99.0%)

Cancer IHC. breast cancer: 1/10 (10.0%); urothelial cancer: 1/11 (9.1%)

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia axoneme (PubMed:36191189). Acts as a regulator of cilium basal body docking and positioning in mono- and multiciliated cells (By similarity). Regulates basal body docking and cilia formation in multiciliated lung cells (By similarity). Regulates kinocilium positioning and stereocilia bundle morphogenesis in the inner ear (By similarity)

Curated tissue note. Expressed in airway epithelial cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VTH2 | HPA portal (live; not the pinned values)

CFAP141 | Unsafe / High RNA

ENSG00000163263

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 143.4 nTPM; lung/ciliated cells; hpa_single_cell_tissue; vital; 11.36 TPM; blood/CD4_STIM; dice_cd4_stim; vital

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 31/176 (17.6%); Glioblastoma Multiforme (validation): 7/58 (12.1%); Breast Invasive Carcinoma (validation): 6/50 (12.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia axoneme, which is required for motile cilia beating

Curated tissue note. Expressed in airway epithelial cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VU69 | HPA portal (live; not the pinned values)

CFAP144P1 | Unsafe / High RNA

ENSG00000164556

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 31.69 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. No exact-ID RNA coverage in the HPA cancer snapshot

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZVS7 | HPA portal (live; not the pinned values)

CFAP20DC | Unsafe / RNA

ENSG00000163689

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. esophagus: 7.7 nTPM (limit 6); esophagus: 7.7 nTPM (baseline 6)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 50/133 (37.6%); Lung Squamous Cell Carcinoma (validation): 24/68 (35.3%); Rectum Adenocarcinoma (validation): 61/207 (29.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZVT6 | HPA portal (live; not the pinned values)

CFAP210 | Unsafe / Protein

ENSG00000154479

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Approved); nasopharynx / respiratory epithelial cells: Medium (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 48/58 (82.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 29/176 (16.5%); Glioblastoma Multiforme (TCGA): 17/141 (12.1%)

Cancer IHC. liver cancer: 6/12 (50.0%); breast cancer: 5/12 (41.7%)

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia axoneme, which is required for motile cilia beating

Curated tissue note. Expressed in airway epithelial cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q0VFZ6 | HPA portal (live; not the pinned values)

CFAP263 | Unsafe / RNA

ENSG00000103021

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.4 nTPM (limit 2); amygdala: 5.6 nTPM (limit 2); basal ganglia: 6.7 nTPM (limit 2); cerebellum: 2.1 nTPM (limit 2); cerebral cortex: 6.4 nTPM (limit 2); choroid plexus: 9.3 nTPM (limit 2); heart muscle: 3.2 nTPM (limit 2); hippocampal formation: 6.2 nTPM (limit 2); hypothalamus: 5.7 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 176/207 (85.0%); Breast Invasive Carcinoma (validation): 42/50 (84.0%)

Cancer IHC. melanoma: 12/12 (100.0%); lung cancer: 5/10 (50.0%)

Biology. Component of centriolar satellites contributing to primary cilium formation (PubMed:25074808). In complex with CFAP263, acts as a regulator of ciliary beating that connects radial spoke 3 (RS3) to the inner dynein arm (IDA) and the nexin-dynein regulatory complex (N-DRC). The complex is positioned parallel to N-DRC and forms a connection between the arch at the base of RS3, the IDA tail and N-DRC (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0I3 | HPA portal (live; not the pinned values)

CFAP298 | Unsafe / Protein

ENSG00000159079

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); adrenal gland / glandular cells: Low (Enhanced); bronchus / respiratory epithelial cells: High (Enhanced); cerebellum / Purkinje cells: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); colon / glandular cells: Medium (Enhanced); duodenum ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. testis cancer: 7/10 (70.0%); colorectal cancer: 6/10 (60.0%)

Biology. Plays a role in motile cilium function, possibly by acting on outer dynein arm assembly (PubMed:24094744). Seems to be important for initiation rather than maintenance of cilium motility (By similarity). Required for correct positioning of the cilium at the apical cell surface, suggesting an additional role in the planar cell polarity (PCP) pathway (By similarity). May suppress canonical Wnt signaling activity (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P57076 | HPA portal (live; not the pinned values)

CFAP299 | Unsafe / Protein

ENSG00000197826

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. nasopharynx / ciliated cells (ciliary rootlets): Low (Approved)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 15/176 (8.5%); Lung Adenocarcinoma (TCGA): 15/497 (3.0%); Ovary Serous Cystadenocarcinoma (validation): 2/81 (2.5%)

Cancer IHC. testis cancer: 8/9 (88.9%); melanoma: 8/12 (66.7%)

Biology. May be involved in spermatogenesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6V702 | HPA portal (live; not the pinned values)

CFAP300 | Unsafe / Protein

ENSG00000137691

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Low (Enhanced); bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (ciliary rootlets): Low (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); nasopharynx / ciliated cells (ciliary rootlets): Low ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 51/105 (48.6%); Glioblastoma Multiforme (validation): 25/58 (43.1%); Ovary Serous Cystadenocarcinoma (TCGA): 141/349 (40.4%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Cilium- and flagellum-specific protein that plays a role in axonemal structure organization and motility. May play a role in outer and inner dynein arm assembly

Curated tissue note. Expressed in nasal epithelial cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BRQ4 | HPA portal (live; not the pinned values)

CFAP418 | Unsafe / Protein

ENSG00000156172

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebral cortex / endothelial cells: Medium (Approved); cerebral cortex / neuronal cells: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Glioblastoma Multiforme (TCGA): 49/141 (34.8%)

Cancer IHC. lymphoma: 5/11 (45.5%); melanoma: 3/10 (30.0%)

Biology. May be involved in photoreceptor outer segment disk morphogenesis (By similarity)

Curated tissue note. Widely expressed, with highest levels in heart and brain. Also expressed in the retina (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96NL8 | HPA portal (live; not the pinned values)

CFAP43 | Unsafe / RNA

ENSG00000197748

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 5.2 nTPM (limit 2); basal ganglia: 9.9 nTPM (limit 2); choroid plexus: 96.9 nTPM (limit 2); hippocampal formation: 9.3 nTPM (limit 2); hypothalamus: 11.9 nTPM (limit 2); kidney: 5.7 nTPM (limit 2); lung: 7.9 nTPM (limit 2); midbrain: 2.7 nTPM (limit 2); pancreas: 2.1 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 23/58 (39.7%); Kidney Renal Papillary Cell Carcinoma (TCGA): 106/282 (37.6%); Uterine Corpus Endometrial Carcinoma (TCGA): 61/176 (34.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Flagellar protein involved in sperm flagellum axoneme organization and function (By similarity). Involved in the regulation of the beating frequency of motile cilia on the epithelial cells of the respiratory tract (By similarity)

Curated tissue note. Testis-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NDM7 | HPA portal (live; not the pinned values)

CFAP44 | Unsafe / Protein

ENSG00000206530

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / neuronal cells: Low (Uncertain); skin 2 / epidermal cells: Low (Uncertain); spleen / cells in red pulp: Low (Uncertain)

Cancer RNA. Glioblastoma Multiforme (validation): 54/58 (93.1%); Pancreatic Adenocarcinoma (validation): 55/80 (68.8%); Rectum Adenocarcinoma (validation): 79/207 (38.2%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Flagellar protein involved in sperm flagellum axoneme organization and function

Curated tissue note. Testis-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96MT7 | HPA portal (live; not the pinned values)

CFAP53 | Unsafe / Protein

ENSG00000172361

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Low (Enhanced); bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); caudate / glial cells: Medium (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 47/58 (81.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 63/176 (35.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 95/282 (33.7%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia axoneme, which is required for motile cilia beating (PubMed:36191189). Regulates motility patterns of both 9+0 and 9+2 motile cilia through differential localization and recruitment of axonemal dynein components (By similarity). Required for centriolar satellite integrity and non-motile cilium assembly (PubMed:26538025). Required for ... [full text in table]

Curated tissue note. Expressed in skin fibroblasts (at protein level) (PubMed:22577226, PubMed:28621423). Expressed in nasal respiratory epithelial cells (at protein level) (PubMed:25504577). Expressed in airway epithelial cells ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96M91 | HPA portal (live; not the pinned values)

CFAP57 | Unsafe / Protein

ENSG00000243710

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); caudate / glial cells: Low (Enhanced); lung / alveolar cells type I: High (Enhanced); lung / macrophages: Medium ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 32/176 (18.2%); Glioblastoma Multiforme (validation): 7/58 (12.1%); Lung Adenocarcinoma (validation): 10/105 (9.5%)

Cancer IHC. colorectal cancer: 2/11 (18.2%); endometrial cancer: 2/11 (18.2%)

Biology. Associates with components of the nexin-dynein regulatory complex (N-DRC), a key regulator of ciliary/flagellar motility, and might act as an inner dynein arm (IDA) hub or linkage

Curated tissue note. Expressed in nasal epithelial cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96MR6 | HPA portal (live; not the pinned values)

CFAP58 | Unsafe / RNA

ENSG00000120051

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3.2 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (validation): 4/58 (6.9%); Uterine Corpus Endometrial Carcinoma (TCGA): 8/176 (4.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 5/283 (1.8%)

Cancer IHC. prostate cancer: 7/12 (58.3%); testis cancer: 2/11 (18.2%)

Biology. Has an essential role in the assembly and organization of the sperm flagellar axoneme (PubMed:32791035). Required for the elongation of the primary cilium and sperm flagellar midpiece via modulation of the Notch signaling pathway (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T655 | HPA portal (live; not the pinned values)

CFAP70 | Unsafe / Protein

ENSG00000156042

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); lung / macrophages: Medium (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 29/58 (50.0%); Ovary Serous Cystadenocarcinoma (validation): 29/81 (35.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 66/282 (23.4%)

Cancer IHC. endometrial cancer: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. Axoneme-binding protein that plays a role in the regulation of ciliary motility and cilium length. Essential for the assembly of the sperm flagellum and the correct shaping of the sperm head, during spermiogenesis.Contributes in part to flagellar biogenesis by stabilizing QRICH2, promoting the cytoplasmic preassembly of CFAP61 and CFAP91, and regulating the manchette localization of axoneme-associated proteins such as IFT88, DNAI1 and ... [full text in table]

Curated tissue note. Expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T0N1 | HPA portal (live; not the pinned values)

CFAP91 | Unsafe / Protein

ENSG00000183833

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Supported); bronchus / ciliated cells (ciliary rootlets): Medium (Supported); bronchus / ciliated cells (tip of cilia): High (Supported)

Cancer RNA. Glioblastoma Multiforme (validation): 40/58 (69.0%); Ovary Serous Cystadenocarcinoma (validation): 27/81 (33.3%); Kidney Renal Clear Cell Carcinoma (validation): 29/100 (29.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Involved in sperm flagellum axonemal organization and function (PubMed:12223483, PubMed:32161152). May regulate cilium motility through its role in the assembly of the axonemal radial spokes (By similarity)

Curated tissue note. Strongly expressed in the liver Widely expressed, but strongly expressed in all spermatogenesis-related tissues, including the testis, the epithelium of cauda and the corpus epididymis, as well as the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z4T9 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CFAP92 | Unsafe / RNA

ENSG00000114656

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.1 nTPM (limit 2); choroid plexus: 4.5 nTPM (limit 2); retina: 2.8 nTPM (limit 2); bone marrow: 2.1 nTPM (baseline 2); choroid plexus: 4.5 nTPM (baseline 2); retina: 2.8 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 182/207 (87.9%); Colon Adenocarcinoma (validation): 425/486 (87.4%); Rectum Adenocarcinoma (TCGA): 34/88 (38.6%)

Cancer IHC. glioma: 3/12 (25.0%); testis cancer: 1/10 (10.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9ULG3 | HPA portal (live; not the pinned values)

CFAP96 | Unsafe / Protein

ENSG00000205129

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cilia axoneme): High (Enhanced); nasopharynx / ciliated cells (tip of cilia): High (Enhanced); skeletal muscle / myocytes: Low (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 50/58 (86.2%); Kidney Renal Clear Cell Carcinoma (validation): 77/100 (77.0%); Breast Invasive Carcinoma (validation): 33/50 (66.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Detected in testis and fetal liver

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A7E2U8 | HPA portal (live; not the pinned values)

CFAP99 | Unsafe / RNA

ENSG00000206113

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.5 nTPM (limit 2); choroid plexus: 2.5 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 4/58 (6.9%); Uterine Corpus Endometrial Carcinoma (TCGA): 11/176 (6.2%); Glioblastoma Multiforme (TCGA): 3/141 (2.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt D6REC4 | HPA portal (live; not the pinned values)

CFHR5 | Unsafe / RNA

ENSG00000134389

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. liver: 162 nTPM (limit 6); liver: 162 nTPM (baseline 2)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 192/231 (83.1%); Liver Hepatocellular Carcinoma (TCGA): 285/362 (78.7%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. liver cancer: 3/12 (25.0%); pancreatic cancer: 1/10 (10.0%)

Biology. Involved in complement regulation. The dimerized forms have avidity for tissue-bound complement fragments and efficiently compete with the physiological complement inhibitor CFH

Curated tissue note. Expressed by the liver and secreted in plasma

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BXR6 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CGB1 | Unsafe / Protein

ENSG00000267631

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. pituitary gland / cells in anterior : High (Approved)

Cancer RNA. Kidney Chromophobe (TCGA): 1/64 (1.6%); Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Ovary Serous Cystadenocarcinoma (TCGA): 4/349 (1.1%)

Cancer IHC. lung cancer: 3/12 (25.0%); endometrial cancer: 1/12 (8.3%)

Biology. Expressed in placenta, testis and pituitary

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NKQ9 | HPA portal (live; not the pinned values)

CGB5 | Unsafe / RNA

ENSG00000189052

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pituitary gland: 8.4 nTPM (limit 2)

Cancer RNA. Pancreatic Adenocarcinoma (TCGA): 84/176 (47.7%); Pancreatic Adenocarcinoma (validation): 19/80 (23.8%); Testicular Germ Cell Tumor (TCGA): 25/133 (18.8%)

Cancer IHC. urothelial cancer: 1/10 (10.0%); breast cancer: 0/10 (0.0%)

Biology. Beta subunit of the human chorionic gonadotropin (hCG). hCG is a complex glycoprotein composed of two glycosylated subunits alpha and beta which are non-covalently associated. The alpha subunit is identical to those in the pituitary gonadotropin hormones (LH, FSH and TSH). The beta subunits are distinct in each of the hormones and confer receptor and biological specificity. Has an essential role in pregnancy and maternal adaptation. ... [full text in table]

Curated tissue note. High expression in the placenta throughout pregnancy

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P0DN86 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CGB8 | Unsafe / RNA

ENSG00000213030

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. pituitary gland: 2.3 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 50/133 (37.6%); Pancreatic Adenocarcinoma (TCGA): 59/176 (33.5%); Pancreatic Adenocarcinoma (validation): 13/80 (16.2%)

Cancer IHC. urothelial cancer: 1/10 (10.0%); breast cancer: 0/10 (0.0%)

Biology. Beta subunit of the human chorionic gonadotropin (hCG). hCG is a complex glycoprotein composed of two glycosylated subunits alpha and beta which are non-covalently associated. The alpha subunit is identical to those in the pituitary gonadotropin hormones (LH, FSH and TSH). The beta subunits are distinct in each of the hormones and confer receptor and biological specificity. Has an essential role in pregnancy and maternal adaptation. ... [full text in table]

Curated tissue note. High expression in the placenta throughout pregnancy

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P0DN86 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CHEK1 | Unsafe / RNA

ENSG00000149554

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 26 nTPM (limit 2); colon: 8 nTPM (limit 6); duodenum: 6.6 nTPM (limit 2); kidney: 2.3 nTPM (limit 2); liver: 2.1 nTPM (limit 2); lymph node: 14.7 nTPM (limit 6); parathyroid gland: 12.8 nTPM (limit 6); rectum: 8.6 nTPM (limit 6); small intestine: 7.3 nTPM (limit 2); smooth muscle: 3.3 ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%); Rectum Adenocarcinoma (validation): 205/207 (99.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Serine/threonine-protein kinase which is required for checkpoint-mediated cell cycle arrest and activation of DNA repair in response to the presence of DNA damage or unreplicated DNA (PubMed:11535615, PubMed:12399544, PubMed:12446774, PubMed:14559997, PubMed:14988723, PubMed:15311285, PubMed:15650047, PubMed:15665856, PubMed:32357935). May also negatively regulate cell cycle progression during unperturbed cell cycles (PubMed:11535615, ... [full text in table]

Curated tissue note. Expressed ubiquitously with the most abundant expression in thymus, testis, small intestine and colon

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14757 | HPA portal (live; not the pinned values)

CHODL | Unsafe / Protein

ENSG00000154645

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / glandular cells: Low (Enhanced); duodenum / glandular cells: Low (Enhanced); rectum / glandular cells: Low (Enhanced); small intestine / glandular cells: Low (Enhanced)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 254/349 (72.8%); Ovary Serous Cystadenocarcinoma (validation): 48/81 (59.3%); Testicular Germ Cell Tumor (TCGA): 61/133 (45.9%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May play a role in the development of the nervous system such as in neurite outgrowth and elongation. May be involved in motor axon growth and guidance

Curated tissue note. Found in spleen, testis, prostate and fetal liver. Expression limited to vascular muscle of testis, smooth muscle of prostate stroma, heart muscle, skeletal muscle, crypts of small intestine, and red pulp of ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H9P2 | HPA portal (live; not the pinned values)

CHRM3 | Unsafe / Protein

ENSG00000133019

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Low (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebral cortex / glial cells: Low (Approved); cerebral cortex / neuronal cells: Medium (Approved); colon / endothelial ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 174/207 (84.1%); Colon Adenocarcinoma (validation): 375/486 (77.2%); Glioblastoma Multiforme (validation): 33/58 (56.9%)

Cancer IHC. renal cancer: 7/10 (70.0%); liver cancer: 4/12 (33.3%)

Biology. The muscarinic acetylcholine receptor mediates various cellular responses, including inhibition of adenylate cyclase, breakdown of phosphoinositides and modulation of potassium channels through the action of G proteins. Primary transducing effect is Pi turnover

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P20309 | HPA portal (live; not the pinned values)

CHRNA5 | Unsafe / Protein

ENSG00000169684

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / glial cells: High (Approved); cerebral cortex / neuronal cells: Medium (Approved); cerebral cortex / neuropil: Medium (Approved); cerebral cortex / synapses: Low (Approved); hippocampus / glial cells: High (Approved); hippocampus / synapses: Low (Approved); smooth muscle / smooth ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 389/486 (80.0%); Pancreatic Adenocarcinoma (validation): 64/80 (80.0%); Testicular Germ Cell Tumor (TCGA): 100/133 (75.2%)

Cancer IHC. skin cancer: 1/12 (8.3%); urothelial cancer: 1/12 (8.3%)

Biology. Component of neuronal acetylcholine receptors (nAChRs) that function as pentameric, ligand-gated cation channels with high calcium permeability among other activities. nAChRs are excitatory neurotrasnmitter receptors formed by a collection of nAChR subunits known to mediate synaptic transmission in the nervous system and the neuromuscular junction. Each nAchR subunit confers differential attributes to channel properties, including ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P30532 | HPA portal (live; not the pinned values)

CHRNA7 | Unsafe / Protein

ENSG00000175344

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 15/80 (18.8%); Stomach Adenocarcinoma (TCGA): 47/346 (13.6%); Colon Adenocarcinoma (validation): 59/486 (12.1%)

Cancer IHC. colorectal cancer: 10/11 (90.9%); prostate cancer: 9/10 (90.0%)

Biology. Component of neuronal acetylcholine receptors (nAChRs) that function as pentameric, ligand-gated cation channels with high calcium permeability among other activities. nAChRs are excitatory neurotrasnmitter receptors formed by a collection of nAChR subunits known to mediate synaptic transmission in the nervous system and the neuromuscular junction. Each nAchR subunit confers differential attributes to channel properties, including ... [full text in table]

Curated tissue note. Expressed in neuronal cells (PubMed:8145738). Expressed in macrophages (at protein level) (PubMed:12508119)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P36544 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CHRNB4 | Unsafe / RNA

ENSG00000117971

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.4 nTPM (limit 2); retina: 2.7 nTPM (limit 2); adrenal gland: 2.4 nTPM (baseline 2); retina: 2.7 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 58/133 (43.6%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 56/283 (19.8%); Lung Squamous Cell Carcinoma (TCGA): 29/489 (5.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of neuronal acetylcholine receptors (nAChRs) that function as pentameric, ligand-gated cation channels with high calcium permeability among other activities. nAChRs are excitatory neurotrasnmitter receptors formed by a collection of nAChR subunits known to mediate synaptic transmission in the nervous system and the neuromuscular junction. Each nAchR subunit confers differential attributes to channel properties, including ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P30926 | HPA portal (live; not the pinned values)

CHURC1 | Unsafe / Protein

ENSG00000258289

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: Medium (Approved); bronchus / ciliated cells (cell body): Medium (Approved); bronchus / ciliated cells (cilia axoneme): Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. breast cancer: 8/11 (72.7%); melanoma: 6/11 (54.5%)

Biology. Transcriptional activator that mediates FGF signaling during neural development (By similarity). Plays a role in the regulation of cell movement (By similarity) Does not bind DNA by itself

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WUH1 | HPA portal (live; not the pinned values)

CILP2 | Unsafe / High RNA

ENSG00000160161

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 159 nTPM; skeletal muscle/fibroblasts; hpa_single_cell_tissue; vital; 5.361 TPM; blood/CD8_STIM; dice_cd8_stim; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Ovary Serous Cystadenocarcinoma (TCGA): 179/349 (51.3%); Breast Invasive Carcinoma (TCGA): 435/1022 (42.6%)

Cancer IHC. liver cancer: 8/12 (66.7%); lung cancer: 3/10 (30.0%)

Biology. May play a role in cartilage scaffolding

Curated tissue note. Expressed in articular chondrocytes but not in knee meniscal cartilage cells (PubMed:12746903). Localizes to the intermediate to deep zone of articular cartilage (PubMed:21880736)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IUL8 | HPA portal (live; not the pinned values)

CIMAP1C | Unsafe / RNA

ENSG00000182950

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.9 nTPM (limit 2); choroid plexus: 2.9 nTPM (baseline 2)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 8/99 (8.1%); Breast Invasive Carcinoma (validation): 4/50 (8.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 20/283 (7.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IXM7 | HPA portal (live; not the pinned values)

CIMAP1D | Unsafe / RNA

ENSG00000181781

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skeletal muscle: 8 nTPM (limit 2); skeletal muscle: 8 nTPM (baseline 2)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 2/99 (2.0%); Kidney Chromophobe (TCGA): 1/64 (1.6%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%)

Cancer IHC. melanoma: 9/12 (75.0%); testis cancer: 7/12 (58.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3SX64 | HPA portal (live; not the pinned values)

CIMIP1 | Unsafe / Protein

ENSG00000124237

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): High (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cell body): High (Enhanced); nasopharynx / ciliated cells (cilia axoneme): Medium (Enhanced); ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 68/176 (38.6%); Lung Squamous Cell Carcinoma (validation): 26/68 (38.2%); Lung Adenocarcinoma (validation): 40/105 (38.1%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Expressed in airway epithelial cells, renal tubular cells, pancreatic acinar cells and epithelial cells of the stomach, duodenum, and gallbladder (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H1P6 | HPA portal (live; not the pinned values)

CIMIP2C | Unsafe / RNA

ENSG00000173557

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 8.5 nTPM (limit 2); hypothalamus: 2.3 nTPM (limit 2)

Cancer RNA. Rectum Adenocarcinoma (TCGA): 51/88 (58.0%); Colon Adenocarcinoma (TCGA): 123/254 (48.4%); Rectum Adenocarcinoma (validation): 86/207 (41.5%)

Cancer IHC. melanoma: 2/12 (16.7%); testis cancer: 1/11 (9.1%)

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia axoneme, which is required for motile cilia beating (PubMed:36191189). Binds to the intra-tubulin interfaces (By similarity)

Curated tissue note. Expressed in airway epithelial cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NJV1 | HPA portal (live; not the pinned values)

CIMIP6 | Unsafe / RNA

ENSG00000177994

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3.8 nTPM (limit 2); choroid plexus: 3.8 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 6/58 (10.3%); Breast Invasive Carcinoma (validation): 3/50 (6.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 8/176 (4.5%)

Cancer IHC. melanoma: 5/12 (41.7%); ovarian cancer: 2/10 (20.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N5S3 | HPA portal (live; not the pinned values)

CIP2A | Unsafe / Protein

ENSG00000163507

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Squamous Cell Carcinoma (validation): 60/68 (88.2%); Pancreatic Adenocarcinoma (validation): 66/80 (82.5%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Acts as an inhibitor of protein phosphatase PP2A (PubMed:17632056). Promotes anchorage-independent cell growth and tumor formation by preventing dephosphorylation of MYC, thereby stabilizing MYC in human malignancies (PubMed:17632056). Together with TOPBP1, plays an essential role in the response to genome instability generated by the presence of acentric chromosome fragments derived from shattered chromosomes within micronuclei ... [full text in table]

Curated tissue note. Expressed at low levels in most of the tissues. Overexpressed in head-and-neck squamous cell carcinomas (HNSCC). Present in liver cancer cells (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TCG1 | HPA portal (live; not the pinned values)

CKAP2L | Unsafe / Protein

ENSG00000169607

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); cerebral cortex / neuronal cells: Low (Uncertain); cerebral cortex / neuropil: Low (Uncertain); colon / endothelial cells: Low (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Colon Adenocarcinoma (validation): 461/486 (94.9%); Rectum Adenocarcinoma (validation): 179/207 (86.5%)

Cancer IHC. ovarian cancer: 6/12 (50.0%); colorectal cancer: 5/11 (45.5%)

Biology. Microtubule-associated protein required for mitotic spindle formation and cell-cycle progression in neural progenitor cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYA6 | HPA portal (live; not the pinned values)

CKS2 | Unsafe / Protein

ENSG00000123975

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: High (Approved); cerebellum ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); lung cancer: 12/12 (100.0%)

Biology. Binds to the catalytic subunit of the cyclin dependent kinases and is essential for their biological function

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P33552 | HPA portal (live; not the pinned values)

CLDN6 | Unsafe / High RNA

ENSG00000184697

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 64.8 nTPM; pancreas/ductal cells; hpa_single_cell_tissue; vital; 0.6714 TPM; blood/CD8_STIM; dice_cd8_stim; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 132/133 (99.2%); Ovary Serous Cystadenocarcinoma (TCGA): 277/349 (79.4%); Ovary Serous Cystadenocarcinoma (validation): 50/81 (61.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays a major role in tight junction-specific obliteration of the intercellular space (Microbial infection) Acts as a receptor for hepatitis C virus (HCV) entry into hepatic cells

Curated tissue note. Expressed in the liver, in peripheral blood mononuclear cells and hepatocarcinoma cell lines

Therapeutic evidence. Clinical outcomes.

UniProt P56747 | Reviewed source 1 | HPA portal (live; not the pinned values)

CLDND2 | Unsafe / RNA

ENSG00000160318

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.4 nTPM (limit 2); amygdala: 5.5 nTPM (limit 2); basal ganglia: 5.5 nTPM (limit 2); bone marrow: 9.5 nTPM (limit 2); cerebellum: 4.9 nTPM (limit 2); cerebral cortex: 6.2 nTPM (limit 2); choroid plexus: 7.3 nTPM (limit 2); duodenum: 4.4 nTPM (limit 2); heart muscle: 3.2 nTPM (limit ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 79/141 (56.0%); Prostate Adenocarcinoma (TCGA): 218/480 (45.4%); Kidney Renal Clear Cell Carcinoma (validation): 45/100 (45.0%)

Cancer IHC. breast cancer: 10/12 (83.3%); cervical cancer: 9/12 (75.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NHS1 | HPA portal (live; not the pinned values)

CLEC12B | Unsafe / RNA

ENSG00000256660

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 5.4 nTPM (limit 2); bone marrow: 5.4 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Rectum Adenocarcinoma (validation): 2/207 (1.0%); Liver Hepatocellular Carcinoma (validation): 2/231 (0.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Inhibitory receptor postulated to negatively regulate immune and non-immune functions (PubMed:17562706, PubMed:34310951). Upon phosphorylation, recruits SH2 domain-containing PTPN6 and PTPN11 phosphatases to its ITIM motif and antagonizes activation signals (PubMed:17562706, PubMed:34310951). Although it inhibits KLRK1/NKG2D-mediated signaling, it does not bind known ligands of KLRK1/NKG2D and therefore is not its inhibitory ... [full text in table]

Curated tissue note. Detected in colon, heart, kidney, liver, lung, mammary gland, ovary, spleen and testis (PubMed:17562706). Expressed in melanocytes (at protein level) (PubMed:34310951)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q2HXU8 | HPA portal (live; not the pinned values)

CLEC18A | Unsafe / RNA

ENSG00000157322

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 15.9 nTPM (limit 2); kidney: 23.2 nTPM (limit 2); choroid plexus: 15.9 nTPM (baseline 2); kidney: 23.2 nTPM (baseline 2)

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 84/100 (84.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 433/521 (83.1%); Glioblastoma Multiforme (TCGA): 38/141 (27.0%)

Cancer IHC. prostate cancer: 5/10 (50.0%); colorectal cancer: 5/12 (41.7%)

Biology. Binds polysaccharides in a Ca(2+)-independent manner with a preferentially binding to fucoidan, beta-glucans and galactans (PubMed:26170455)

Curated tissue note. Dectected in all cell lines tested and in peripheral blood cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A5D8T8 | HPA portal (live; not the pinned values)

CLEC2D | Unsafe / Protein

ENSG00000069493

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Squamous Cell Carcinoma (validation): 63/68 (92.6%); Breast Invasive Carcinoma (validation): 46/50 (92.0%)

Cancer IHC. lymphoma: 11/12 (91.7%); colorectal cancer: 9/12 (75.0%)

Biology. Receptor for KLRB1 that protects target cells against natural killer cell-mediated lysis (PubMed:16339513, PubMed:20843815). Inhibits osteoclast formation (PubMed:14753741, PubMed:15123656). Inhibits bone resorption (PubMed:14753741). Modulates the release of interferon-gamma (PubMed:15104121). Binds high molecular weight sulfated glycosaminoglycans (PubMed:15123656)

Curated tissue note. Detected in peripheral blood leukocytes, osteoblasts, lymph node, thymus and spleen. Isoform 1, isoform 2 and isoform 4 are expressed in T- and B-lymphocytes, and at lower levels in NK cells. They are also ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UHP7 | HPA portal (live; not the pinned values)

CLEC4C | Unsafe / Protein

ENSG00000198178

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 8/133 (6.0%); Breast Invasive Carcinoma (validation): 3/50 (6.0%); Bladder Urothelial Carcinoma (TCGA): 4/169 (2.4%)

Cancer IHC. liver cancer: 12/12 (100.0%); renal cancer: 12/12 (100.0%)

Biology. Lectin-type cell surface receptor which may play a role in antigen capturing by dendritic cells (PubMed:11748283, PubMed:21880719, PubMed:25995448). Specifically recognizes non-sialylated galactose-terminated biantennary glycans containing the trisaccharide epitope Gal(beta1-3/4)GlcNAc(beta1-2)Man (PubMed:21880719, PubMed:25995448). Binds to serum IgG (PubMed:25995448). Efficiently targets ligand into antigen-processing and ... [full text in table]

Curated tissue note. Expressed in plasmacytoid dendritic cells (PDCs). Constitutively expressed in immature monocyte-derived dendritic cells (iMDDC) and is significantly down-regulated upon maturation with LPS but not with TNF

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WTT0 | HPA portal (live; not the pinned values)

CLGN | Unsafe / RNA

ENSG00000153132

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 7.9 nTPM (limit 2); heart muscle: 57.4 nTPM (limit 12); hypothalamus: 4.8 nTPM (limit 2); pituitary gland: 10.9 nTPM (limit 2); retina: 16.7 nTPM (limit 2)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 447/480 (93.1%); Breast Invasive Carcinoma (validation): 39/50 (78.0%); Ovary Serous Cystadenocarcinoma (TCGA): 218/349 (62.5%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Functions during spermatogenesis as a chaperone for a range of client proteins that are important for sperm adhesion onto the egg zona pellucida and for subsequent penetration of the zona pellucida. Required for normal sperm migration from the uterus into the oviduct. Required for normal male fertility. Binds calcium ions (By similarity)

Curated tissue note. Detected in testis (at protein level). Detected in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14967 | HPA portal (live; not the pinned values)

CLHC1 | Unsafe / RNA

ENSG00000162994

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.7 nTPM (limit 2); bone marrow: 2.1 nTPM (limit 2); cerebellum: 2.3 nTPM (limit 2); cerebral cortex: 2.5 nTPM (limit 2); choroid plexus: 4.2 nTPM (limit 2); heart muscle: 2.3 nTPM (limit 2); kidney: 3.5 nTPM (limit 2); lung: 2.4 nTPM (limit 2); pancreas: 2.5 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 165/207 (79.7%); Colon Adenocarcinoma (validation): 351/486 (72.2%)

Cancer IHC. liver cancer: 4/11 (36.4%); ovarian cancer: 4/12 (33.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NHS4 | HPA portal (live; not the pinned values)

CLIP3 | Unsafe / RNA

ENSG00000105270

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 15.8 nTPM (limit 10); adrenal gland: 13.1 nTPM (limit 2); amygdala: 151.5 nTPM (limit 2); basal ganglia: 132.8 nTPM (limit 2); cerebellum: 215.1 nTPM (limit 2); cerebral cortex: 213.9 nTPM (limit 2); choroid plexus: 29.7 nTPM (limit 2); colon: 72.7 nTPM (limit 6); duodenum: 6.3 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Ovary Serous Cystadenocarcinoma (TCGA): 326/349 (93.4%)

Cancer IHC. glioma: 5/11 (45.5%); liver cancer: 1/12 (8.3%)

Biology. Functions as a cytoplasmic linker protein. Involved in TGN-endosome dynamics. May modulate the cellular compartmentalization of AKT kinase family and promote its cell membrane localization, thereby playing a role in glucose transport in adipocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96DZ5 | HPA portal (live; not the pinned values)

CLNK | Unsafe / Protein

ENSG00000109684

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / cells in tubules: Low (Uncertain); lymph node / non-germinal center cells: High (Uncertain)

Cancer RNA. Kidney Chromophobe (TCGA): 57/64 (89.1%); Uterine Corpus Endometrial Carcinoma (TCGA): 9/176 (5.1%); Pancreatic Adenocarcinoma (validation): 4/80 (5.0%)

Cancer IHC. testis cancer: 3/11 (27.3%); lung cancer: 2/9 (22.2%)

Biology. An adapter protein which plays a role in the regulation of immunoreceptor signaling, including PLC-gamma-mediated B-cell antigen receptor (BCR) signaling and FC-epsilon R1-mediated mast cell degranulation (By similarity). Together with FGR, it acts as a negative regulator of natural killer cell-activating receptors and inhibits interferon-gamma production (By similarity). Acts as a positive regulator of both T-cell receptor and natural ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z7G1 | HPA portal (live; not the pinned values)

CLPB | Unsafe / Protein

ENSG00000162129

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 484/486 (99.6%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Functions as a regulatory ATPase and participates in secretion/protein trafficking process. Has ATP-dependent protein disaggregase activity and is required to maintain the solubility of key mitochondrial proteins (PubMed:32573439, PubMed:34115842, PubMed:35247700, PubMed:36170828, PubMed:36745679). Involved in mitochondrial-mediated antiviral innate immunity, activates RIG-I-mediated signal transduction and production of IFNB1 and ... [full text in table]

Curated tissue note. Widely expressed (at protein level) (PubMed:25597511). Expressed in fetal, as well as in adult tissues, with highest levels in adult brain, including thalamus, hippocampus, occipital cortex and parietal ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H078 | HPA portal (live; not the pinned values)

CLPSL1 | Unsafe / RNA

ENSG00000204140

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. hippocampal formation: 2.8 nTPM (limit 2); pancreas: 173.3 nTPM (limit 12)

Cancer RNA. Breast Invasive Carcinoma (TCGA): 559/1022 (54.7%); Uterine Corpus Endometrial Carcinoma (TCGA): 59/176 (33.5%); Ovary Serous Cystadenocarcinoma (TCGA): 73/349 (20.9%)

Cancer IHC. renal cancer: 6/12 (50.0%); endometrial cancer: 2/12 (16.7%)

Biology. Exclusively expressed in epididymis, in the corpus region

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A2RUU4 | HPA portal (live; not the pinned values)

CLPSL2 | Unsafe / Protein

ENSG00000196748

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Medium (Approved); colon / glandular cells: Low (Approved); kidney / cells in tubules: Low (Approved); lung / ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 450/1022 (44.0%); Lung Adenocarcinoma (TCGA): 52/497 (10.5%); Uterine Corpus Endometrial Carcinoma (TCGA): 12/176 (6.8%)

Cancer IHC. prostate cancer: 4/11 (36.4%); breast cancer: 3/11 (27.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6UWE3 | HPA portal (live; not the pinned values)

CLRN1 | Unsafe / RNA

ENSG00000163646

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.8 nTPM (limit 2); retina: 12.4 nTPM (limit 2); adrenal gland: 4.8 nTPM (baseline 2); retina: 12.4 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (TCGA): 2/141 (1.4%); Bladder Urothelial Carcinoma (TCGA): 2/169 (1.2%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May have a role in the excitatory ribbon synapse junctions between hair cells and cochlear ganglion cells and presumably also in analogous synapses within the retina

Curated tissue note. Widely expressed. Found in the retina

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P58418 | HPA portal (live; not the pinned values)

CLSPN | Unsafe / RNA

ENSG00000092853

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 11.1 nTPM (limit 2); retina: 6.6 nTPM (limit 2); bone marrow: 11.1 nTPM (baseline 2); retina: 6.6 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Colon Adenocarcinoma (validation): 447/486 (92.0%); Rectum Adenocarcinoma (validation): 169/207 (81.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Required for checkpoint mediated cell cycle arrest in response to inhibition of DNA replication or to DNA damage induced by both ionizing and UV irradiation (PubMed:12766152, PubMed:15190204, PubMed:15707391, PubMed:16123041). Adapter protein which binds to BRCA1 and the checkpoint kinase CHEK1 and facilitates the ATR-dependent phosphorylation of both proteins (PubMed:12766152, PubMed:15096610, PubMed:15707391, PubMed:16123041). Also ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HAW4 | HPA portal (live; not the pinned values)

CLUL1 | Unsafe / Protein

ENSG00000079101

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: High (Enhanced)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 387/480 (80.6%); Ovary Serous Cystadenocarcinoma (TCGA): 202/349 (57.9%); Kidney Renal Clear Cell Carcinoma (validation): 53/100 (53.0%)

Cancer IHC. endometrial cancer: 4/12 (33.3%); prostate cancer: 1/12 (8.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15846 | HPA portal (live; not the pinned values)

CMTM1 | Unsafe / RNA

ENSG00000089505

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. cerebellum: 2.3 nTPM (limit 2); cerebellum: 2.3 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 48/50 (96.0%); Colon Adenocarcinoma (validation): 460/486 (94.7%); Rectum Adenocarcinoma (validation): 191/207 (92.3%)

Cancer IHC. testis cancer: 5/10 (50.0%); prostate cancer: 5/11 (45.5%)

Biology. Highly expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZ96 | HPA portal (live; not the pinned values)

CMTM2 | Unsafe / High RNA

ENSG00000140932

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 1249 nTPM; fallopian tube/neutrophils; hpa_single_cell_tissue; vital; 19.11 TPM; blood/MONOCYTES; dice_monocytes; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 3/50 (6.0%); Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Highly expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TAZ6 | HPA portal (live; not the pinned values)

CNBD1 | Unsafe / High RNA

ENSG00000176571

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 40.1 nTPM; brain/inhibitory neurons; hpa_single_cell_tissue; vital; 0.4798 TPM; blood/MONOCYTES; dice_monocytes; vital

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Lung Adenocarcinoma (TCGA): 1/497 (0.2%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. prostate cancer: 2/12 (16.7%); testis cancer: 2/12 (16.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NA66 | HPA portal (live; not the pinned values)

CNBD2 | Unsafe / Protein

ENSG00000149646

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 114/141 (80.9%); Prostate Adenocarcinoma (TCGA): 388/480 (80.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 100/176 (56.8%)

Cancer IHC. liver cancer: 12/12 (100.0%); stomach cancer: 12/12 (100.0%)

Biology. Essential for male fertility. Plays an important role in spermatogenesis and regulates sperm motility by controlling the development of the flagellar bending of sperm

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96M20 | HPA portal (live; not the pinned values)

CNKSR3 | Unsafe / Protein

ENSG00000153721

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 222/231 (96.1%); Colon Adenocarcinoma (validation): 461/486 (94.9%); Kidney Renal Clear Cell Carcinoma (validation): 93/100 (93.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); lung cancer: 12/12 (100.0%)

Biology. Involved in transepithelial sodium transport. Regulates aldosterone-induced and epithelial sodium channel (ENaC)-mediated sodium transport through regulation of ENaC cell surface expression. Acts as a scaffold protein coordinating the assembly of an ENaC-regulatory complex (ERC)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6P9H4 | HPA portal (live; not the pinned values)

CNST | Unsafe / Protein

ENSG00000162852

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. skin cancer: 9/11 (81.8%); prostate cancer: 6/11 (54.5%)

Biology. Required for targeting of connexins to the plasma membrane

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PJW8 | HPA portal (live; not the pinned values)

CNTD1 | Unsafe / High RNA

ENSG00000176563

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 43.2 nTPM; lung/ciliated cells; hpa_single_cell_tissue; vital; 14.2 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%); Breast Invasive Carcinoma (validation): 37/50 (74.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 40/283 (14.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays a role in the different steps of crossover formation during meiotic recombination. Participates in the crossover differentiation step of crossover-specific recombination intermediates through its interaction with PRR19. In addition, stimulates crossover formation through the interactions with RFC3 and RFC4 and simultaneously regulates cell-cycle progression through interactions with CDC34 and subsequent ubiquitination of WEE1. ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N815 | HPA portal (live; not the pinned values)

CNTLN | Unsafe / Protein

ENSG00000044459

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / cells in ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Pancreatic Adenocarcinoma (validation): 55/80 (68.8%)

Cancer IHC. prostate cancer: 12/12 (100.0%); breast cancer: 11/12 (91.7%)

Biology. Required for centrosome cohesion and recruitment of CEP68 to centrosomes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NXG0 | HPA portal (live; not the pinned values)

CNTN4 | Unsafe / RNA

ENSG00000144619

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 4.7 nTPM (limit 2); cerebral cortex: 7.5 nTPM (limit 2); heart muscle: 2.4 nTPM (limit 2); kidney: 4 nTPM (limit 2); liver: 2.8 nTPM (limit 2); lung: 2.3 nTPM (limit 2); parathyroid gland: 21.8 nTPM (limit 6); retina: 19.2 nTPM (limit 2); small intestine: 3.9 nTPM (limit 2); smooth ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 36/50 (72.0%); Pancreatic Adenocarcinoma (validation): 41/80 (51.2%); Thyroid Carcinoma (TCGA): 190/495 (38.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Contactins mediate cell surface interactions during nervous system development. Has some neurite outgrowth-promoting activity. May be involved in synaptogenesis

Curated tissue note. Mainly expressed in brain. Highly expressed in cerebellum and weakly expressed in corpus callosum, caudate nucleus, amygdala and spinal cord. Also expressed in testis, pancreas, thyroid, uterus, small ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IWV2 | HPA portal (live; not the pinned values)

COL10A1 | Unsafe / RNA

ENSG00000123500

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. lung: 2.8 nTPM (limit 2); pancreas: 3.2 nTPM (limit 2); lung: 2.8 nTPM (baseline 2); pancreas: 3.2 nTPM (baseline 2)

Cancer RNA. Lung Adenocarcinoma (validation): 104/105 (99.0%); Lung Squamous Cell Carcinoma (validation): 67/68 (98.5%); Pancreatic Adenocarcinoma (validation): 76/80 (95.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Type X collagen is a product of hypertrophic chondrocytes and has been localized to presumptive mineralization zones of hyaline cartilage

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q03692 | HPA portal (live; not the pinned values)

COL20A1 | Unsafe / RNA

ENSG00000101203

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 7.5 nTPM (limit 2); basal ganglia: 4.3 nTPM (limit 2); cerebral cortex: 9 nTPM (limit 2); hippocampal formation: 6 nTPM (limit 2); hypothalamus: 3.2 nTPM (limit 2); midbrain: 5 nTPM (limit 2); spinal cord: 4.2 nTPM (limit 2); amygdala: 7.5 nTPM (baseline 2); basal ganglia: 4.3 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 85/141 (60.3%); Glioblastoma Multiforme (validation): 29/58 (50.0%); Skin Cuteneous Melanoma (TCGA): 8/99 (8.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Probable collagen protein

Curated tissue note. High expression in heart, lung, liver, skeletal muscle, kidney, pancreas, spleen, testis, ovary, subthalamic nucleus and fetal liver. Weak expression in other tissues tested

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P218 | HPA portal (live; not the pinned values)

CORO7 | Unsafe / Protein

ENSG00000262246

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); colon / glandular cells: High (Uncertain); duodenum / glandular cells: High (Uncertain); kidney / cells in glomeruli: Medium (Uncertain); kidney / cells in tubules: Low (Uncertain); lung ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Rectum Adenocarcinoma (TCGA): 88/88 (100.0%); Colon Adenocarcinoma (TCGA): 252/254 (99.2%)

Cancer IHC. colorectal cancer: 4/12 (33.3%); liver cancer: 2/10 (20.0%)

Biology. F-actin regulator involved in anterograde Golgi to endosome transport: upon ubiquitination via 'Lys-33'-linked ubiquitin chains by the BCR(KLHL20) E3 ubiquitin ligase complex, interacts with EPS15 and localizes to the trans-Golgi network, where it promotes actin polymerization, thereby facilitating post-Golgi trafficking. May play a role in the maintenance of the Golgi apparatus morphology

Curated tissue note. Widely expressed. Expressed in the spleen, peripheral leukocytes, testes, brain, thymus and small intestine

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P57737 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

COX6B2 | Unsafe / RNA

ENSG00000160471

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. colon: 6.5 nTPM (limit 6); colon: 6.5 nTPM (baseline 6)

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 142/283 (50.2%); Pancreatic Adenocarcinoma (TCGA): 85/176 (48.3%); Pancreatic Adenocarcinoma (validation): 34/80 (42.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of the cytochrome c oxidase, the last enzyme in the mitochondrial electron transport chain which drives oxidative phosphorylation. The respiratory chain contains 3 multisubunit complexes succinate dehydrogenase (complex II, CII), ubiquinol-cytochrome c oxidoreductase (cytochrome b-c1 complex, complex III, CIII) and cytochrome c oxidase (complex IV, CIV), that cooperate to transfer electrons derived from NADH and succinate to ... [full text in table]

Curated tissue note. Testis specific. Weak expression in thymus and heart. Expressed in cancer cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6YFQ2 | HPA portal (live; not the pinned values)

COX8C | Unsafe / Protein

ENSG00000187581

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Approved); cerebral cortex / endothelial cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); colon / endothelial cells: Medium (Approved); colon / peripheral nerve/ganglion: Medium (Approved); duodenum / glandular cells: Low (Approved); ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 27/133 (20.3%); Ovary Serous Cystadenocarcinoma (TCGA): 30/349 (8.6%); Ovary Serous Cystadenocarcinoma (validation): 4/81 (4.9%)

Cancer IHC. glioma: 11/12 (91.7%); melanoma: 10/11 (90.9%)

Biology. Component of the cytochrome c oxidase, the last enzyme in the mitochondrial electron transport chain which drives oxidative phosphorylation. The respiratory chain contains 3 multisubunit complexes succinate dehydrogenase (complex II, CII), ubiquinol-cytochrome c oxidoreductase (cytochrome b-c1 complex, complex III, CIII) and cytochrome c oxidase (complex IV, CIV), that cooperate to transfer electrons derived from NADH and succinate to ... [full text in table]

Curated tissue note. It is not yet known where COX8C is expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z4L0 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CPA5 | Unsafe / High RNA

ENSG00000158525

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 106.6 nTPM; pbmc/t-cells; hpa_single_cell_tissue; vital; 82.54 TPM; blood/T-reg; hpa_immune_cell_samples; vital

Cancer RNA. Pancreatic Adenocarcinoma (TCGA): 6/176 (3.4%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 8/283 (2.8%); Bladder Urothelial Carcinoma (TCGA): 4/169 (2.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Expression is very low or not detectable

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WXQ8 | HPA portal (live; not the pinned values)

CPT1A | Unsafe / Protein

ENSG00000110090

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: Low (Supported); bronchus / basal cells: High (Supported); bronchus / ciliated cells (cell body): High (Supported); bronchus / goblet cells: High (Supported); caudate / glial cells: Low (Supported); caudate / ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. liver cancer: 10/11 (90.9%); ovarian cancer: 9/10 (90.0%)

Biology. Catalyzes the transfer of the acyl group of long-chain fatty acid-CoA conjugates onto carnitine, an essential step for the mitochondrial uptake of long-chain fatty acids and their subsequent beta-oxidation in the mitochondrion (PubMed:11350182, PubMed:14517221, PubMed:16651524, PubMed:9691089). Also possesses a lysine succinyltransferase activity that can regulate enzymatic activity of substrate proteins such as ENO1 and metabolism ... [full text in table]

Curated tissue note. Strong expression in kidney and heart, and lower in liver and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P50416 | HPA portal (live; not the pinned values)

CPT1C | Unsafe / Protein

ENSG00000169169

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); hippocampus / neuronal cells: Medium (Approved); kidney / cells in tubules: Low (Approved); salivary gland / glandular cells: Low (Approved); smooth ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 140/141 (99.3%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 42/50 (84.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Palmitoyl thioesterase specifically expressed in the endoplasmic reticulum of neurons. Modulates the trafficking of the glutamate receptor, AMPAR, to plasma membrane through depalmitoylation of GRIA1 (PubMed:30135643). Also regulates AMPR trafficking through the regulation of SACM1L phosphatidylinositol-3-phosphatase activity by interaction in a malonyl-CoA dependent manner (By similarity). Binds malonyl-CoA and couples malonyl-CoA to ... [full text in table]

Curated tissue note. Expressed predominantly in brain and testis. Expressed in motor neurons

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TCG5 | HPA portal (live; not the pinned values)

CRACD | Unsafe / Protein

ENSG00000109265

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 200/207 (96.6%); Colon Adenocarcinoma (validation): 468/486 (96.3%)

Cancer IHC. melanoma: 6/8 (75.0%); pancreatic cancer: 6/9 (66.7%)

Biology. Involved in epithelial cell integrity by acting on the maintenance of the actin cytoskeleton. Positively regulates the actin polymerization, by inhibiting the interaction of actin-capping proteins with actin

Curated tissue note. Expressed in intestinal epithelial cells (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZU35 | HPA portal (live; not the pinned values)

CRB1 | Unsafe / Protein

ENSG00000134376

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / outer plexiform layer: Low (Approved); retina / photoreceptor cells: Medium (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Glioblastoma Multiforme (TCGA): 123/141 (87.2%); Skin Cuteneous Melanoma (TCGA): 6/99 (6.1%)

Cancer IHC. liver cancer: 1/12 (8.3%); pancreatic cancer: 1/12 (8.3%)

Biology. Plays a role in photoreceptor morphogenesis in the retina (By similarity). May maintain cell polarization and adhesion (By similarity)

Curated tissue note. Preferential expression in retina, also expressed in brain, testis, fetal brain and fetal eye (PubMed:15914641). Expressed at the outer limiting membrane and apical to adherens junctions in the retina ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P82279 | HPA portal (live; not the pinned values)

CRCP | Unsafe / Protein

ENSG00000241258

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Low (Approved); bronchus / ciliated cells (cell body): Medium (Approved); bronchus / ciliated cells (cilia axoneme): High (Approved); bronchus / ciliated cells (ciliary rootlets): High (Approved); bronchus / ciliated ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Ovary Serous Cystadenocarcinoma (TCGA): 349/349 (100.0%)

Cancer IHC. breast cancer: 4/11 (36.4%); lung cancer: 4/11 (36.4%)

Biology. DNA-dependent RNA polymerase catalyzes the transcription of DNA into RNA using the four ribonucleoside triphosphates as substrates (PubMed:20413673, PubMed:33558764, PubMed:34675218). Specific peripheric component of RNA polymerase III (Pol III) which synthesizes small non-coding RNAs including 5S rRNA, snRNAs, tRNAs and miRNAs from at least 500 distinct genomic loci. With POLR3H/RPC8 forms a mobile stalk that protrudes from Pol III ... [full text in table]

Curated tissue note. Ubiquitous. Most prevalent in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75575 | HPA portal (live; not the pinned values)

CREM | Unsafe / Protein

ENSG00000095794

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); cerebellum / Purkinje cells - cytoplasm/membrane: Low (Enhanced); colon / goblet cells: Medium (Enhanced); colon / mucosal lymphoid ... [full text in table]

Cancer RNA. Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%)

Cancer IHC. pancreatic cancer: 4/11 (36.4%); prostate cancer: 3/10 (30.0%)

Biology. Transcriptional regulator that binds the cAMP response element (CRE), a sequence present in many viral and cellular promoters. Isoforms are either transcriptional activators or repressors. Plays a role in spermatogenesis and is involved in spermatid maturation (PubMed:10373550) May play a role in the regulation of the circadian clock: acts as a transcriptional repressor of the core circadian component PER1 by directly binding to cAMP ... [full text in table]

Curated tissue note. Expressed in testes (round spermatids) (at protein level). Isoform 14 is the major activator form in testes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q03060 | HPA portal (live; not the pinned values)

CRH | Unsafe / RNA

ENSG00000147571

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 5.7 nTPM (limit 2); basal ganglia: 3.4 nTPM (limit 2); cerebral cortex: 11.5 nTPM (limit 2); hippocampal formation: 3.9 nTPM (limit 2); hypothalamus: 46 nTPM (limit 2); amygdala: 5.7 nTPM (baseline 2); basal ganglia: 3.4 nTPM (baseline 2); cerebral cortex: 11.5 nTPM (baseline 2); ... [full text in table]

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 43/169 (25.4%); Kidney Chromophobe (TCGA): 15/64 (23.4%); Pancreatic Adenocarcinoma (TCGA): 17/176 (9.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Hormone that plays a central role in whole body adaptation to stress (By similarity). Released by the hypothalamus primarily in response to physical or psychological stress and acts by binding to CRH receptor CRHR1, and to a lower extent, CRHR2 (PubMed:18801728, PubMed:3003585, PubMed:32004469). CRH-dependent signaling is a primary mediator of the neuroendocrine, autonomic (fight-or-flight) and behavioral responses to stress, acting as ... [full text in table]

Curated tissue note. Produced by the hypothalamus and placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P06850 | HPA portal (live; not the pinned values)

CRIP1 | Unsafe / Protein

ENSG00000213145

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / goblet cells: Medium (Approved); cerebral cortex / glial cells: Low (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: Low (Approved); duodenum / glandular ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. breast cancer: 8/10 (80.0%); ovarian cancer: 9/12 (75.0%)

Biology. Seems to have a role in zinc absorption and may function as an intracellular zinc transport protein

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P50238 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CRISP2 | Unsafe / RNA

ENSG00000124490

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 16.5 nTPM (limit 12); choroid plexus: 4.1 nTPM (limit 2)

Cancer RNA. Pancreatic Adenocarcinoma (TCGA): 20/176 (11.4%); Breast Invasive Carcinoma (validation): 4/50 (8.0%); Lung Adenocarcinoma (validation): 7/105 (6.7%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May regulate some ion channels' activity and thereby regulate calcium fluxes during sperm capacitation

Curated tissue note. Testis and epididymis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P16562 | HPA portal (live; not the pinned values)

CRYBA4 | Unsafe / Protein

ENSG00000196431

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. eye / lens fiber cells: High (Supported)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 11/133 (8.3%); Uterine Corpus Endometrial Carcinoma (TCGA): 8/176 (4.5%); Glioblastoma Multiforme (validation): 1/58 (1.7%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Crystallins are the dominant structural components of the vertebrate eye lens

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P53673 | HPA portal (live; not the pinned values)

CRYGC | Unsafe / Protein

ENSG00000163254

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. eye / lens fiber cells: High (Supported)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 14/349 (4.0%); Ovary Serous Cystadenocarcinoma (validation): 3/81 (3.7%); Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%)

Cancer IHC. liver cancer: 1/11 (9.1%); renal cancer: 1/12 (8.3%)

Biology. Crystallins are the dominant structural components of the vertebrate eye lens

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P07315 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CSH1 | Unsafe / Protein

ENSG00000136488

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. pituitary gland / cells in anterior : High (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 26/133 (19.5%); Breast Invasive Carcinoma (validation): 1/50 (2.0%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Produced only during pregnancy and is involved in stimulating lactation, fetal growth and metabolism. Does not interact with GHR but only activates PRLR through zinc-induced dimerization

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P0DML2 | HPA portal (live; not the pinned values)

CSH2 | Unsafe / RNA

ENSG00000213218

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. smooth muscle: 14.6 nTPM (limit 6)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 15/133 (11.3%); Breast Invasive Carcinoma (validation): 1/50 (2.0%); Rectum Adenocarcinoma (validation): 2/207 (1.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Produced only during pregnancy and is involved in stimulating lactation, fetal growth and metabolism. Does not interact with GHR but only activates PRLR through zinc-induced dimerization

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P0DML3 | HPA portal (live; not the pinned values)

CSHL1 | Unsafe / Protein

ENSG00000204414

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. pituitary gland / cells in anterior : High (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May be a novel gestational hormone required to compensate for absence of other members of the GH/CS cluster during gestation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14406 | HPA portal (live; not the pinned values)

CSMD2 | Unsafe / RNA

ENSG00000121904

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.5 nTPM (limit 2); cerebral cortex: 2.3 nTPM (limit 2); hypothalamus: 2.4 nTPM (limit 2); basal ganglia: 2.5 nTPM (baseline 2); cerebral cortex: 2.3 nTPM (baseline 2); hypothalamus: 2.4 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 50/58 (86.2%); Pancreatic Adenocarcinoma (validation): 15/80 (18.8%); Glioblastoma Multiforme (TCGA): 24/141 (17.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Weakly expressed in most tissues, except in brain. Expressed at intermediate level in brain, including cerebellum, substantia nigra, hippocampus and fetal brain. Overexpressed in some head and neck cancer cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z408 | HPA portal (live; not the pinned values)

CSMD3 | Unsafe / RNA

ENSG00000164796

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.7 nTPM (limit 2); hypothalamus: 2.1 nTPM (limit 2); cerebral cortex: 2.7 nTPM (baseline 2); hypothalamus: 2.1 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 30/58 (51.7%); Breast Invasive Carcinoma (validation): 6/50 (12.0%); Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%)

Cancer IHC. testis cancer: 11/12 (91.7%); endometrial cancer: 9/12 (75.0%)

Biology. Involved in dendrite development

Curated tissue note. Weakly expressed in most tissues, except in brain. Expressed at intermediate level in brain, including cerebellum, substantia nigra, thalamus, spinal cord, hippocampus and fetal brain. Also expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z407 | HPA portal (live; not the pinned values)

CSN2 | Unsafe / High RNA

ENSG00000135222

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 683.2 nTPM; breast/macrophages; hpa_single_cell_tissue; vital; 0.3195 TPM; blood/TH1; dice_th1; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 3/50 (6.0%); Liver Hepatocellular Carcinoma (validation): 4/231 (1.7%); Ovary Serous Cystadenocarcinoma (validation): 1/81 (1.2%)

Cancer IHC. colorectal cancer: 7/12 (58.3%); testis cancer: 5/12 (41.7%)

Biology. Important role in determination of the surface properties of the casein micelles

Curated tissue note. Mammary gland specific. Secreted in milk

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P05814 | HPA portal (live; not the pinned values)

CSN3 | Unsafe / RNA

ENSG00000171209

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. salivary gland: 65.7 nTPM (limit 24)

Cancer RNA. Breast Invasive Carcinoma (validation): 9/50 (18.0%); Lung Squamous Cell Carcinoma (validation): 5/68 (7.4%); Breast Invasive Carcinoma (TCGA): 42/1022 (4.1%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Kappa-casein stabilizes micelle formation, preventing casein precipitation in milk

Curated tissue note. Mammary gland specific. Secreted in milk

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P07498 | HPA portal (live; not the pinned values)

CSPP1 | Unsafe / RNA

ENSG00000104218

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.8 nTPM (limit 2); amygdala: 4 nTPM (limit 2); basal ganglia: 5 nTPM (limit 2); cerebellum: 7 nTPM (limit 2); cerebral cortex: 4 nTPM (limit 2); choroid plexus: 3.8 nTPM (limit 2); duodenum: 2.2 nTPM (limit 2); heart muscle: 3.6 nTPM (limit 2); hippocampal formation: 4.8 nTPM (limit ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 49/50 (98.0%); Pancreatic Adenocarcinoma (validation): 77/80 (96.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May play a role in cell-cycle-dependent microtubule organization

Curated tissue note. Expressed in adult and fetal brain with enrichment in the cerebellum. Detected in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q1MSJ5 | HPA portal (live; not the pinned values)

CT45A1 | Unsafe / Protein

ENSG00000268940

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. skin 2 / epidermal cells: Low (Supported)

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 98/489 (20.0%); Ovary Serous Cystadenocarcinoma (TCGA): 61/349 (17.5%); Lung Squamous Cell Carcinoma (validation): 10/68 (14.7%)

Cancer IHC. ovarian cancer: 4/10 (40.0%); testis cancer: 3/12 (25.0%)

Biology. Testis specific. Expressed in cancer cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5HYN5 | HPA portal (live; not the pinned values)

CT45A3 | Unsafe / Protein

ENSG00000269096

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. heart muscle / cardiomyocytes: Low (Supported)

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 94/489 (19.2%); Ovary Serous Cystadenocarcinoma (TCGA): 64/349 (18.3%); Lung Squamous Cell Carcinoma (validation): 11/68 (16.2%)

Cancer IHC. ovarian cancer: 4/10 (40.0%); testis cancer: 3/12 (25.0%)

Biology. Testis specific. Expressed in cancer cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NHU0 | HPA portal (live; not the pinned values)

CT45A5 | Unsafe / RNA

ENSG00000228836

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 4.8 nTPM (limit 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 82/349 (23.5%); Lung Squamous Cell Carcinoma (TCGA): 114/489 (23.3%); Lung Squamous Cell Carcinoma (validation): 11/68 (16.2%)

Cancer IHC. ovarian cancer: 4/10 (40.0%); testis cancer: 3/12 (25.0%)

Biology. Testis specific. Expressed in cancer cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P0DMU7 | UniProt P0DMU8 | UniProt P0DMV0 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CT45A6 | Unsafe / RNA

ENSG00000278289

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 3.9 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 6/133 (4.5%); Ovary Serous Cystadenocarcinoma (TCGA): 14/349 (4.0%); Lung Squamous Cell Carcinoma (TCGA): 16/489 (3.3%)

Cancer IHC. ovarian cancer: 4/10 (40.0%); testis cancer: 3/12 (25.0%)

Biology. Testis specific. Expressed in cancer cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P0DMU7 | UniProt P0DMU8 | UniProt P0DMV0 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

CTDSPL2 | Unsafe / Protein

ENSG00000137770

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); cerebral cortex / glial cells: Low (Approved); cerebral cortex / neuropil: Low (Approved); colon / glandular cells: Medium (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. skin cancer: 10/12 (83.3%); cervical cancer: 8/11 (72.7%)

Biology. Serine/threonine phosphatase that promotes gluconeogenesis by dephosphorylating FOXO1 and FOXO3A, promoting their nuclear retention and enhancing their transcriptional activity on PCK1 and G6PC1, key enzymes in hepatic gluconeogenesis (PubMed:28851713). Essential for proper embryonic skeletal development by regulating chondrocyte survival through dephosphorylation of FOXO3 (By similarity). By maintaining FOXO3 in its active, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q05D32 | HPA portal (live; not the pinned values)

CTNNA2 | Unsafe / Protein

ENSG00000066032

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / glial cells: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); cerebral cortex / neuropil: Medium (Enhanced); hippocampus / neuronal cells: Medium (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Glioblastoma Multiforme (TCGA): 137/141 (97.2%); Kidney Chromophobe (TCGA): 34/64 (53.1%)

Cancer IHC. ovarian cancer: 3/10 (30.0%); colorectal cancer: 3/11 (27.3%)

Biology. May function as a linker between cadherin adhesion receptors and the cytoskeleton to regulate cell-cell adhesion and differentiation in the nervous system (By similarity). Required for proper regulation of cortical neuronal migration and neurite growth (PubMed:30013181). It acts as a negative regulator of Arp2/3 complex activity and Arp2/3-mediated actin polymerization (PubMed:30013181). It thereby suppresses excessive actin branching ... [full text in table]

Curated tissue note. Expressed in neural tissues, with strongest expression in fetal and adult brain. Expressed in the developing cortical plate and marginal zone of 20-week-old human fetal brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P26232 | HPA portal (live; not the pinned values)

CXCL11 | Unsafe / RNA

ENSG00000169248

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 9 nTPM (limit 2); cerebral cortex: 5.4 nTPM (limit 2); hippocampal formation: 3.1 nTPM (limit 2); hypothalamus: 3.6 nTPM (limit 2); liver: 2.3 nTPM (limit 2); lung: 5.8 nTPM (limit 2); lymph node: 11.6 nTPM (limit 6); midbrain: 2.7 nTPM (limit 2); pancreas: 8.6 nTPM (limit 2); spinal ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 42/50 (84.0%); Lung Squamous Cell Carcinoma (validation): 56/68 (82.4%); Testicular Germ Cell Tumor (TCGA): 89/133 (66.9%)

Cancer IHC. colorectal cancer: 9/9 (100.0%); renal cancer: 11/12 (91.7%)

Biology. Chemotactic for interleukin-activated T-cells but not unstimulated T-cells, neutrophils or monocytes. Induces calcium release in activated T-cells. Binds to CXCR3. May play an important role in CNS diseases which involve T-cell recruitment. May play a role in skin immune responses

Curated tissue note. High levels in peripheral blood leukocytes, pancreas and liver astrocytes. Moderate levels in thymus, spleen and lung. Low levels in placenta, prostate and small intestine. Also found in epidermal basal layer ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14625 | HPA portal (live; not the pinned values)

CXorf65 | Unsafe / High RNA

ENSG00000204165

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 3.7 nTPM; pbmc/t-cells; hpa_single_cell_tissue; vital; 198.8 TPM; blood/NK; dice_nk; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 10/50 (20.0%); Skin Cuteneous Melanoma (TCGA): 4/99 (4.0%); Liver Hepatocellular Carcinoma (validation): 9/231 (3.9%)

Cancer IHC. testis cancer: 6/11 (54.5%); breast cancer: 1/12 (8.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NEN9 | HPA portal (live; not the pinned values)

CYB5RL | Unsafe / Protein

ENSG00000215883

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / Purkinje cells: Medium (Approved); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 40/50 (80.0%); Glioblastoma Multiforme (validation): 45/58 (77.6%); Rectum Adenocarcinoma (validation): 119/207 (57.5%)

Cancer IHC. ovarian cancer: 2/9 (22.2%); glioma: 2/12 (16.7%)

Biology. Putative NADH-cytochrome b5 reductases family protein. May participate in fatty acid desaturation and elongation, cholesterol biosynthesis, drug metabolism, and, in erythrocyte, methemoglobin reduction

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6IPT4 | HPA portal (live; not the pinned values)

CYP11B1 | Unsafe / Protein

ENSG00000160882

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / cells in zona fasciculata: High (Supported); adrenal gland / cells in zona glomerulosa: High (Supported); adrenal gland / cells in zona reticularis: High (Supported); adrenal gland / glandular cells: High (Supported)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 1/282 (0.4%); Lung Adenocarcinoma (TCGA): 1/497 (0.2%)

Cancer IHC. breast cancer: 0/9 (0.0%); cervical cancer: 0/7 (0.0%)

Biology. A cytochrome P450 monooxygenase mainly involved in the biosynthesis of adrenal corticoids (PubMed:12530636, PubMed:1518866, PubMed:1775135, PubMed:18215163, PubMed:23322723, PubMed:23685396, PubMed:29277707). Catalyzes a variety of reactions that are essential for many species, including detoxification, defense, and the formation of endogenous chemicals like steroid hormones. Steroid 11beta, 18- and 19-hydroxylase with preferred ... [full text in table]

Curated tissue note. Expressed in the zona fasciculata/reticularis of the adrenal cortex

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P15538 | HPA portal (live; not the pinned values)

CYP11B2 | Unsafe / Protein

ENSG00000179142

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / cells in zona fasciculata: High (Supported); adrenal gland / cells in zona glomerulosa: High (Supported); adrenal gland / cells in zona reticularis: High (Supported); adrenal gland / glandular cells: High (Supported)

Cancer RNA. Liver Hepatocellular Carcinoma (TCGA): 3/362 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. breast cancer: 0/9 (0.0%); cervical cancer: 0/7 (0.0%)

Biology. A cytochrome P450 monooxygenase that catalyzes the biosynthesis of aldosterone, the main mineralocorticoid in the human body responsible for salt and water homeostasis, thus involved in blood pressure regulation, arterial hypertension, and the development of heart failure (PubMed:11856349, PubMed:12530636, PubMed:1518866, PubMed:15356073, PubMed:1594605, PubMed:1775135, PubMed:22446688, PubMed:23322723, PubMed:9814482, PubMed:9814506). ... [full text in table]

Curated tissue note. Expressed sporadically in the zona glomerulosa (zG) of the adrenal cortex (conventional zonation), as well as in aldosterone-producing cell clusters (APCCs) composed of morphological zG cells in contact with ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P19099 | HPA portal (live; not the pinned values)

CYP19A1 | Unsafe / High RNA

ENSG00000137869

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 33.4 nTPM; skeletal muscle/fibroblasts; hpa_single_cell_tissue; vital; 0.7468 TPM; blood/neutrophil; hpa_immune_cell_samples; vital

Cancer RNA. Glioblastoma Multiforme (validation): 13/58 (22.4%); Colon Adenocarcinoma (validation): 62/486 (12.8%); Rectum Adenocarcinoma (validation): 23/207 (11.1%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. A cytochrome P450 monooxygenase that catalyzes the conversion of C19 androgens, androst-4-ene-3,17-dione (androstenedione) and testosterone to the C18 estrogens, estrone and estradiol, respectively (PubMed:27702664, PubMed:2848247). Catalyzes three successive oxidations of C19 androgens: two conventional oxidations at C19 yielding 19-hydroxy and 19-oxo/19-aldehyde derivatives, followed by a third oxidative aromatization step that ... [full text in table]

Curated tissue note. Widely expressed, including in adult and fetal brain, placenta, skin fibroblasts, adipose tissue and gonads

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P11511 | HPA portal (live; not the pinned values)

CYP2C8 | Unsafe / Protein

ENSG00000138115

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / enterocytes: Low (Enhanced); liver / hepatocytes: High (Enhanced); small intestine / enterocytes: Low (Enhanced)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 214/231 (92.6%); Liver Hepatocellular Carcinoma (TCGA): 313/362 (86.5%); Breast Invasive Carcinoma (validation): 18/50 (36.0%)

Cancer IHC. liver cancer: 2/10 (20.0%); breast cancer: 0/11 (0.0%)

Biology. A cytochrome P450 monooxygenase involved in the metabolism of various endogenous substrates, including fatty acids, steroid hormones and vitamins (PubMed:11093772, PubMed:14559847, PubMed:15766564, PubMed:19965576, PubMed:7574697). Mechanistically, uses molecular oxygen inserting one oxygen atom into a substrate, and reducing the second into a water molecule, with two electrons provided by NADPH via cytochrome P450 reductase ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P10632 | HPA portal (live; not the pinned values)

CYTH3 | Unsafe / Protein

ENSG00000008256

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Skin Cuteneous Melanoma (TCGA): 99/99 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. melanoma: 12/12 (100.0%); glioma: 6/12 (50.0%)

Biology. Promotes guanine-nucleotide exchange on ARF1 and ARF6. Promotes the activation of ARF factors through replacement of GDP with GTP. Plays a role in the epithelial polarization (By similarity)

Curated tissue note. Almost absent from liver, thymus and peripheral blood lymphocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43739 | HPA portal (live; not the pinned values)

DAW1 | Unsafe / Protein

ENSG00000123977

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): High (Approved); bronchus / ciliated cells (cilia axoneme): High (Approved); bronchus / ciliated cells (ciliary rootlets): High (Approved); bronchus / ciliated cells (tip of cilia): High (Approved); nasopharynx / ciliated cells (cell body): High (Approved); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 55/497 (11.1%); Lung Adenocarcinoma (validation): 9/105 (8.6%); Kidney Renal Papillary Cell Carcinoma (TCGA): 23/282 (8.2%)

Cancer IHC. prostate cancer: 11/11 (100.0%); pancreatic cancer: 11/12 (91.7%)

Biology. Required for axonemal dynein assembly and ciliary motility in ciliated organs, including Kupffer's vesicle, during embryogenesis (PubMed:36074124). Facilitates the onset of robust cilia motility during development (PubMed:36074124)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N136 | HPA portal (live; not the pinned values)

DAZ4 | Unsafe / RNA

ENSG00000205916

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. stomach: 25.2 nTPM (limit 8)

Cancer RNA. Stomach Adenocarcinoma (TCGA): 46/346 (13.3%); Pancreatic Adenocarcinoma (validation): 8/80 (10.0%); Glioblastoma Multiforme (TCGA): 5/141 (3.5%)

Cancer IHC. liver cancer: 1/12 (8.3%); testis cancer: 1/12 (8.3%)

Biology. RNA-binding protein that plays an essential role in spermatogenesis. May act by binding to the 3'-UTR of mRNAs and regulating their translation

Curated tissue note. Testis-specific. Expression restricted to premeiotic germ cells, particularly in spermatogonia (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86SG3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

DBF4 | Unsafe / High RNA

ENSG00000006634

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 120.1 nTPM; adipose tissue/t-cells; hpa_single_cell_tissue; vital; 113.2 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 78/80 (97.5%); Breast Invasive Carcinoma (validation): 44/50 (88.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Regulatory subunit for CDC7 which activates its kinase activity thereby playing a central role in DNA replication and cell proliferation. Required for progression of S phase. The complex CDC7-DBF4A selectively phosphorylates MCM2 subunit at 'Ser-40' and 'Ser-53' and then is involved in regulating the initiation of DNA replication during cell cycle

Curated tissue note. Highly expressed in testis and thymus. Expressed also in most cancer cells lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UBU7 | HPA portal (live; not the pinned values)

DCAF12L1 | Unsafe / Protein

ENSG00000198889

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / neuronal cells: Medium (Approved); colon / glandular cells: Medium ... [full text in table]

Cancer RNA. Kidney Renal Papillary Cell Carcinoma (TCGA): 68/282 (24.1%); Kidney Renal Clear Cell Carcinoma (validation): 19/100 (19.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 22/176 (12.5%)

Cancer IHC. breast cancer: 11/11 (100.0%); colorectal cancer: 11/12 (91.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VU92 | HPA portal (live; not the pinned values)

DCAF8L1 | Unsafe / Protein

ENSG00000226372

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced); nasopharynx / respiratory epithelial cells: Low (Enhanced)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 11/231 (4.8%); Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Liver Hepatocellular Carcinoma (TCGA): 3/362 (0.8%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NGE4 | HPA portal (live; not the pinned values)

DCHS2 | Unsafe / RNA

ENSG00000197410

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.2 nTPM (limit 2); basal ganglia: 2.2 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 28/58 (48.3%); Pancreatic Adenocarcinoma (validation): 16/80 (20.0%); Breast Invasive Carcinoma (validation): 2/50 (4.0%)

Cancer IHC. skin cancer: 11/12 (91.7%); renal cancer: 10/12 (83.3%)

Biology. Calcium-dependent cell-adhesion protein

Curated tissue note. Cerebral cortex and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6V1P9 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

DCST1 | Unsafe / RNA

ENSG00000163357

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.5 nTPM (limit 2); retina: 2.5 nTPM (baseline 2)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 4/231 (1.7%); Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 4/283 (1.4%)

Cancer IHC. glioma: 9/10 (90.0%); colorectal cancer: 6/10 (60.0%)

Biology. E3 ubiquitin-protein ligase which mediates 'Lys-48'-linked ubiquitination of STAT2 and induces its proteasomal degradation thereby negatively regulating type-I-interferon signaling. Essential sperm cell-surface protein required for sperm-egg fusion and fertilization (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T197 | HPA portal (live; not the pinned values)

DCUN1D4 | Unsafe / Protein

ENSG00000109184

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%)

Cancer IHC. testis cancer: 9/11 (81.8%); renal cancer: 8/10 (80.0%)

Biology. Contributes to the neddylation of all cullins by transferring NEDD8 from N-terminally acetylated NEDD8-conjugating E2s enzyme to different cullin C-terminal domain-RBX complexes which are necessary for the activation of cullin-RING E3 ubiquitin ligases (CRLs)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92564 | HPA portal (live; not the pinned values)

DCX | Unsafe / Protein

ENSG00000077279

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); caudate / synapses: Low (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); cerebral cortex / neuronal projections: Low (Enhanced); colon / glandular cells: Low (Enhanced); hippocampus ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 44/58 (75.9%); Glioblastoma Multiforme (TCGA): 66/141 (46.8%); Breast Invasive Carcinoma (validation): 9/50 (18.0%)

Cancer IHC. glioma: 3/11 (27.3%); pancreatic cancer: 2/11 (18.2%)

Biology. Microtubule-associated protein required for initial steps of neuronal dispersion and cortex lamination during cerebral cortex development. May act by competing with the putative neuronal protein kinase DCLK1 in binding to a target protein. May in that way participate in a signaling pathway that is crucial for neuronal interaction before and during migration, possibly as part of a calcium ion-dependent signal transduction pathway. May ... [full text in table]

Curated tissue note. Highly expressed in neuronal cells of fetal brain (in the majority of cells of the cortical plate, intermediate zone and ventricular zone), but not expressed in other fetal tissues. In the adult, highly ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43602 | HPA portal (live; not the pinned values)

DDHD1 | Unsafe / Protein

ENSG00000100523

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); cerebellum / cells in granular layer: Medium (Enhanced); cerebellum / Purkinje cells: Medium (Enhanced); cerebral cortex / neuropil: Medium (Enhanced); colon / glandular cells: Low (Enhanced); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. melanoma: 4/12 (33.3%); testis cancer: 4/12 (33.3%)

Biology. Phospholipase A1 (PLA1) that hydrolyzes ester bonds at the sn-1 position of glycerophospholipids producing a free fatty acid and a lysophospholipid (Probable) (PubMed:20359546, PubMed:22922100). Prefers phosphatidate (1,2-diacyl-sn-glycero-3-phosphate, PA) as substrate in vitro, but can efficiently hydrolyze phosphatidylinositol (1,2-diacyl-sn-glycero-3-phospho-(1D-myo-inositol), PI), as well as a range of other glycerophospholipid ... [full text in table]

Curated tissue note. Highly expressed in testis. Also expressed in brain, spleen and lung. Only expressed in cerebellum in fetal brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEL9 | HPA portal (live; not the pinned values)

DDIAS | Unsafe / Protein

ENSG00000165490

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); colon / endothelial cells: Medium (Approved); colon / glandular cells: Medium (Approved); duodenum / glandular cells: Medium (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 459/486 (94.4%); Rectum Adenocarcinoma (validation): 189/207 (91.3%); Lung Squamous Cell Carcinoma (validation): 59/68 (86.8%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. May be an anti-apoptotic protein involved in DNA repair or cell survival

Curated tissue note. Highly expressed in colorectal and lung cancer tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IXT1 | HPA portal (live; not the pinned values)

DDX11 | Unsafe / Protein

ENSG00000013573

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Supported); adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: Low (Supported); bronchus / respiratory epithelial cells: Medium (Supported); caudate / glial cells: Medium (Supported); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. testis cancer: 8/10 (80.0%); breast cancer: 6/11 (54.5%)

Biology. DNA-dependent ATPase and ATP-dependent DNA helicase that participates in various functions in genomic stability, including DNA replication, DNA repair and heterochromatin organization as well as in ribosomal RNA synthesis (PubMed:10648783, PubMed:21854770, PubMed:23797032, PubMed:26089203, PubMed:26503245). Its double-stranded DNA helicase activity requires either a minimal 5'-single-stranded tail length of approximately 15 nt (flap ... [full text in table]

Curated tissue note. Expressed in melanoma cells. Not detected in epidermal melanocytes of normal skin (at protein level) (PubMed:23116066). Highly expressed in spleen, B-cells, thymus, testis, ovary, small intestine and pancreas ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96FC9 | HPA portal (live; not the pinned values)

DDX20 | Unsafe / Protein

ENSG00000064703

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced); colon / glandular cells: Low (Enhanced); colon / peripheral nerve/ganglion: Low (Enhanced); duodenum / glandular cells: Low (Enhanced); esophagus / squamous epithelial cells: Low (Enhanced); lymph node / germinal center cells: Low (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 49/50 (98.0%); Pancreatic Adenocarcinoma (validation): 76/80 (95.0%)

Cancer IHC. testis cancer: 8/12 (66.7%); colorectal cancer: 5/12 (41.7%)

Biology. The SMN complex catalyzes the assembly of small nuclear ribonucleoproteins (snRNPs), the building blocks of the spliceosome, and thereby plays an important role in the splicing of cellular pre-mRNAs. Most spliceosomal snRNPs contain a common set of Sm proteins SNRPB, SNRPD1, SNRPD2, SNRPD3, SNRPE, SNRPF and SNRPG that assemble in a heptameric protein ring on the Sm site of the small nuclear RNA to form the core snRNP (Sm core). In the ... [full text in table]

Curated tissue note. Ubiquitous

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UHI6 | HPA portal (live; not the pinned values)

DDX25 | Unsafe / Protein

ENSG00000109832

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / glial cells: High (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); cerebral cortex / neuronal projections: Medium (Enhanced); hypothalamus / glial cells: Medium (Enhanced); hypothalamus / neuronal cells: Low (Enhanced); hypothalamus / neuronal projections: High ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 42/58 (72.4%); Glioblastoma Multiforme (TCGA): 71/141 (50.4%); Testicular Germ Cell Tumor (TCGA): 41/133 (30.8%)

Cancer IHC. liver cancer: 1/10 (10.0%); prostate cancer: 1/12 (8.3%)

Biology. ATP-dependent RNA helicase. Required for mRNA export and translation regulation during spermatid development (By similarity)

Curated tissue note. Highly expressed in the Leydig and germ cells of the testis and weakly expressed in the pituitary and hypothalamus

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UHL0 | HPA portal (live; not the pinned values)

DDX43 | Unsafe / Protein

ENSG00000080007

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. lung / macrophages: Medium (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 97/133 (72.9%); Kidney Renal Clear Cell Carcinoma (validation): 12/100 (12.0%); Lung Adenocarcinoma (validation): 12/105 (11.4%)

Cancer IHC. testis cancer: 9/12 (75.0%); liver cancer: 3/12 (25.0%)

Biology. Expressed in testis. Expressed in many tumors of various histological types at a level that is 100-fold higher than the level observed in normal tissues except testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NXZ2 | HPA portal (live; not the pinned values)

DDX47 | Unsafe / Protein

ENSG00000213782

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / Purkinje cells: High (Approved); cerebral cortex / endothelial cells: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. skin cancer: 11/12 (91.7%); melanoma: 8/9 (88.9%)

Biology. Required for efficient ribosome biogenesis (By similarity). May have a role in mRNA splicing (PubMed:16963496). Involved in apoptosis (PubMed:15977068)

Curated tissue note. Expressed in skin, lung and breast. Also expressed in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0S4 | HPA portal (live; not the pinned values)

DEFB119 | Unsafe / RNA

ENSG00000180483

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 150.8 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 22/133 (16.5%); Glioblastoma Multiforme (validation): 3/58 (5.2%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. stomach cancer: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. Has antibacterial activity

Curated tissue note. Abundant expression in the male reproductive tract only. Abundant expressed in testis and the caput region of epididymis, but low in the corpus region

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N690 | HPA portal (live; not the pinned values)

DEPDC1 | Unsafe / Protein

ENSG00000024526

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. 82.9 nTPM; tongue/basal keratinocytes; hpa_single_cell_tissue; protected; 1.8 TPM; blood/Plasmablast; hpa_immune_monaco; vital

Cancer RNA. Colon Adenocarcinoma (validation): 465/486 (95.7%); Glioblastoma Multiforme (validation): 53/58 (91.4%); Rectum Adenocarcinoma (validation): 174/207 (84.1%)

Cancer IHC. urothelial cancer: 9/10 (90.0%); melanoma: 8/11 (72.7%)

Biology. May be involved in transcriptional regulation as a transcriptional corepressor. The DEPDC1A-ZNF224 complex may play a critical role in bladder carcinogenesis by repressing the transcription of the A20 gene, leading to transport of NF-KB protein into the nucleus, resulting in suppression of apoptosis of bladder cancer cells

Curated tissue note. Expressed in testis. Up-regulated in bladder cancer cells (at protein level)

Therapeutic evidence. Trial registration only.

UniProt Q5TB30 | Trial/source record | HPA portal (live; not the pinned values)

DEPDC1B | Unsafe / Protein

ENSG00000035499

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: Low (Approved); duodenum / ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 228/254 (89.8%); Colon Adenocarcinoma (validation): 435/486 (89.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 244/283 (86.2%)

Cancer IHC. liver cancer: 7/10 (70.0%); endometrial cancer: 5/12 (41.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WUY9 | HPA portal (live; not the pinned values)

DEPDC7 | Unsafe / RNA

ENSG00000121690

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.4 nTPM (limit 2); cerebral cortex: 2.1 nTPM (limit 2); choroid plexus: 2.8 nTPM (limit 2); duodenum: 23.9 nTPM (limit 2); hippocampal formation: 3.3 nTPM (limit 2); kidney: 18.8 nTPM (limit 2); liver: 71.8 nTPM (limit 2); midbrain: 2.8 nTPM (limit 2); pancreas: 3.7 nTPM (limit 2); ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 219/231 (94.8%); Kidney Renal Clear Cell Carcinoma (validation): 86/100 (86.0%); Liver Hepatocellular Carcinoma (TCGA): 304/362 (84.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Expressed in liver

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96QD5 | HPA portal (live; not the pinned values)

DEUP1 | Unsafe / High RNA

ENSG00000165325

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 38.3 nTPM; lung/ciliated cells; hpa_single_cell_tissue; vital; 1 TPM; blood/myeloid DC; hpa_immune_monaco; vital

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 6/176 (3.4%); Lung Squamous Cell Carcinoma (validation): 2/68 (2.9%); Ovary Serous Cystadenocarcinoma (TCGA): 10/349 (2.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Key structural component of the deuterosome, a structure that promotes de novo centriole amplification in multiciliated cells. Deuterosome-mediated centriole amplification occurs in terminally differentiated multiciliated cells and can generate more than 100 centrioles. Probably sufficient for the specification and formation of the deuterosome inner core. Interacts with CEP152 and recruits PLK4 to activate centriole biogenesis (By ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q05D60 | HPA portal (live; not the pinned values)

DGKH | Unsafe / Protein

ENSG00000102780

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); breast cancer: 10/11 (90.9%)

Biology. Diacylglycerol kinase that converts diacylglycerol/DAG into phosphatidic acid/phosphatidate/PA and regulates the respective levels of these two bioactive lipids (PubMed:12810723, PubMed:23949095). Thereby, acts as a central switch between the signaling pathways activated by these second messengers with different cellular targets and opposite effects in numerous biological processes (Probable) (PubMed:12810723, PubMed:23949095). Plays a ... [full text in table]

Curated tissue note. Expressed only in testis, kidney and colon Ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86XP1 | HPA portal (live; not the pinned values)

DHH | Unsafe / High RNA

ENSG00000139549

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 56.9 nTPM; vascular/schwann cells; hpa_single_cell_tissue; vital; 1.014 TPM; blood/NK; dice_nk; vital

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 4/99 (4.0%); Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Precursor of desert hedgehog, a morphogen that activates the smoothened signaling pathway, and which is essential for a variety of patterning events during development (By similarity). The C-terminal part of the precursor displays an autoproteolysis and a cholesterol transferase activity, resulting (1) in the cleavage of the full-length protein into two parts, desert hedgehog protein N-product and C-product and (2) covalent attachment ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43323 | HPA portal (live; not the pinned values)

DHX9 | Unsafe / Protein

ENSG00000135829

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: Medium (Supported); caudate / glial cells: High (Supported); caudate / neuronal cells: High (Supported); ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Colon Adenocarcinoma (validation): 486/486 (100.0%); Ovary Serous Cystadenocarcinoma (TCGA): 349/349 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Multifunctional ATP-dependent nucleic acid helicase that unwinds DNA and RNA in a 3' to 5' direction and that plays important roles in many processes, such as DNA replication, transcriptional activation, post-transcriptional RNA regulation, mRNA translation and RNA-mediated gene silencing (PubMed:11416126, PubMed:12711669, PubMed:15355351, PubMed:16680162, PubMed:17531811, PubMed:20669935, PubMed:21561811, PubMed:24049074, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q08211 | HPA portal (live; not the pinned values)

DIAPH3 | Unsafe / RNA

ENSG00000139734

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 7 nTPM (limit 2); smooth muscle: 2.5 nTPM (limit 2); bone marrow: 7 nTPM (baseline 2); smooth muscle: 2.5 nTPM (baseline 2)

Cancer RNA. Colon Adenocarcinoma (validation): 478/486 (98.4%); Rectum Adenocarcinoma (validation): 196/207 (94.7%); Glioblastoma Multiforme (validation): 53/58 (91.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Actin nucleation and elongation factor required for the assembly of F-actin structures, such as actin cables and stress fibers. Required for cytokinesis, stress fiber formation and transcriptional activation of the serum response factor. Binds to GTP-bound form of Rho and to profilin: acts in a Rho-dependent manner to recruit profilin to the membrane, where it promotes actin polymerization. DFR proteins couple Rho and Src tyrosine ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NSV4 | HPA portal (live; not the pinned values)

DLGAP5 | Unsafe / Protein

ENSG00000126787

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: High (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); cerebral cortex / neuropil: Low (Enhanced); duodenum / endocrine cells: Medium (Enhanced); esophagus / squamous epithelial cells: Low (Enhanced); kidney / cells in glomeruli: Low (Enhanced); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 483/486 (99.4%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 279/283 (98.6%); Glioblastoma Multiforme (validation): 57/58 (98.3%)

Cancer IHC. testis cancer: 11/12 (91.7%); cervical cancer: 8/11 (72.7%)

Biology. Potential cell cycle regulator that may play a role in carcinogenesis of cancer cells. Mitotic phosphoprotein regulated by the ubiquitin-proteasome pathway. Key regulator of adherens junction integrity and differentiation that may be involved in CDH1-mediated adhesion and signaling in epithelial cells

Curated tissue note. Abundantly expressed in fetal liver. Expressed at lower levels in bone marrow, testis, colon, and placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15398 | HPA portal (live; not the pinned values)

DMC1 | Unsafe / Protein

ENSG00000100206

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. heart muscle / cardiomyocytes: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 46/133 (34.6%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 85/283 (30.0%); Breast Invasive Carcinoma (validation): 9/50 (18.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Homologous DNA recombinase that catalyzes strand exchange during meiotic recombination (PubMed:15164066, PubMed:21307306, PubMed:26976601). Specifically required for meiotic recombination, facilitating the search and repair of DNA using the homolog rather than the sister chromatid, and can tolerate mismatches in heteroduplex DNA, unlike the higher-fidelity RAD51 (PubMed:15164066, PubMed:26976601)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14565 | HPA portal (live; not the pinned values)

DMP1 | Unsafe / Protein

ENSG00000152592

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in molecular layer: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 1/58 (1.7%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Kidney Renal Clear Cell Carcinoma (TCGA): 4/521 (0.8%)

Cancer IHC. pancreatic cancer: 12/12 (100.0%); colorectal cancer: 11/11 (100.0%)

Biology. May have a dual function during osteoblast differentiation. In the nucleus of undifferentiated osteoblasts, unphosphorylated form acts as a transcriptional component for activation of osteoblast-specific genes like osteocalcin. During the osteoblast to osteocyte transition phase it is phosphorylated and exported into the extracellular matrix, where it regulates nucleation of hydroxyapatite

Curated tissue note. Expressed in tooth particularly in odontoblast, ameloblast and cementoblast

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13316 | HPA portal (live; not the pinned values)

DMRTC1 | Unsafe / RNA

ENSG00000269502

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 3.2 nTPM (limit 2); retina: 2.3 nTPM (limit 2); cerebral cortex: 3.2 nTPM (baseline 2); retina: 2.3 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 4/50 (8.0%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Stomach Adenocarcinoma (TCGA): 1/346 (0.3%)

Cancer IHC. urothelial cancer: 2/11 (18.2%); pancreatic cancer: 1/8 (12.5%)

Biology. Predominantly expressed in kidney, pancreas, ovary and testis. Detected in brain and in many other tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5HYR2 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

DMRTC1B | Unsafe / RNA

ENSG00000184911

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.4 nTPM (limit 2); amygdala: 21 nTPM (limit 2); basal ganglia: 28.2 nTPM (limit 2); cerebellum: 9.4 nTPM (limit 2); cerebral cortex: 23.2 nTPM (limit 2); choroid plexus: 4.3 nTPM (limit 2); hippocampal formation: 17.3 nTPM (limit 2); hypothalamus: 27 nTPM (limit 2); midbrain: 9.2 ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 164/495 (33.1%); Glioblastoma Multiforme (TCGA): 42/141 (29.8%); Breast Invasive Carcinoma (validation): 13/50 (26.0%)

Cancer IHC. colorectal cancer: 4/12 (33.3%); urothelial cancer: 3/11 (27.3%)

Biology. Predominantly expressed in kidney, pancreas, ovary and testis. Detected in brain and in many other tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5HYR2 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

DNAAF1 | Unsafe / Protein

ENSG00000154099

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): High (Enhanced); bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 453/480 (94.4%); Uterine Corpus Endometrial Carcinoma (TCGA): 161/176 (91.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 87/282 (30.9%)

Cancer IHC. ovarian cancer: 2/11 (18.2%); breast cancer: 0/11 (0.0%)

Biology. Cilium-specific protein required for the stability of the ciliary architecture. Plays a role in cytoplasmic preassembly of dynein arms. Involved in regulation of microtubule-based cilia and actin-based brush border microvilli

Curated tissue note. Mainly expressed in trachea and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEP3 | HPA portal (live; not the pinned values)

DNAAF10 | Unsafe / Protein

ENSG00000243667

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Lung Adenocarcinoma (validation): 104/105 (99.0%); Rectum Adenocarcinoma (validation): 204/207 (98.6%)

Cancer IHC. prostate cancer: 9/9 (100.0%); pancreatic cancer: 2/11 (18.2%)

Biology. Key assembly factor specifically required for the stability of axonemal dynein heavy chains in cytoplasm

Curated tissue note. Widely expressed with the highest expression in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96MX6 | HPA portal (live; not the pinned values)

DNAAF4 | Unsafe / Protein

ENSG00000256061

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in molecular layer: Medium ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 427/495 (86.3%); Kidney Renal Papillary Cell Carcinoma (TCGA): 205/282 (72.7%); Glioblastoma Multiforme (TCGA): 99/141 (70.2%)

Cancer IHC. colorectal cancer: 11/12 (91.7%); pancreatic cancer: 10/12 (83.3%)

Biology. Axonemal dynein assembly factor required for ciliary motility. Involved in neuronal migration during development of the cerebral neocortex. May regulate the stability and proteasomal degradation of the estrogen receptors that play an important role in neuronal differentiation, survival and plasticity

Curated tissue note. Expressed in several tissues, including brain, lung, kidney and testis. In brain localizes to a fraction of cortical neurons and white matter glial cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WXU2 | HPA portal (live; not the pinned values)

DNAAF6 | Unsafe / Protein

ENSG00000080572

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): High (Enhanced); bronchus / ciliated cells (ciliary rootlets): High (Enhanced); bronchus / goblet cells: High (Enhanced)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 21/176 (11.9%); Glioblastoma Multiforme (validation): 2/58 (3.4%); Lung Adenocarcinoma (TCGA): 14/497 (2.8%)

Cancer IHC. endometrial cancer: 1/12 (8.3%); breast cancer: 0/10 (0.0%)

Biology. Plays a role in cytoplasmic pre-assembly of axonemal dynein

Curated tissue note. Expressed in testis, small intestine, prostate, adrenal gland, spleen, lung, bladder, breast and ovary. Expressed in ciliated epithelial cells (PubMed:28176794)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NQM4 | HPA portal (live; not the pinned values)

DNAAF8 | Unsafe / Protein

ENSG00000166246

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); kidney / proximal tubules (cell body): Low (Enhanced); nasopharynx / ciliated cells (cell body): Medium (Enhanced); nasopharynx / ciliated cells (ciliary rootlets): Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 24/50 (48.0%); Breast Invasive Carcinoma (TCGA): 465/1022 (45.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 125/282 (44.3%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. In cyliated cells, dynein axonemal particle-specific protein required for deployment of ODA to the axoneme. Interacts with outer dynein arm (ODA) subunits

Curated tissue note. Expressed in respiratory ciliated cells (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYS4 | HPA portal (live; not the pinned values)

DNAH10 | Unsafe / Protein

ENSG00000197653

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); choroid plexus / ependymal cells: Medium (Enhanced); kidney / cells in tubules: Low (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 7/50 (14.0%); Glioblastoma Multiforme (validation): 1/58 (1.7%); Uterine Corpus Endometrial Carcinoma (TCGA): 3/176 (1.7%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Force generating protein of respiratory cilia. Produces force towards the minus ends of microtubules. Dynein has ATPase activity; the force-producing power stroke is thought to occur on release of ADP. Involved in sperm motility; implicated in sperm flagellar assembly (PubMed:34237282). Probable inner arm dynein heavy chain

Curated tissue note. Expressed primarily in trachea and testis, 2 tissues containing axonemal structures. Also expressed in brain but not in adult heart

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IVF4 | HPA portal (live; not the pinned values)

DNAH11 | Unsafe / Protein

ENSG00000105877

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Approved); bronchus / ciliated cells (ciliary rootlets): Medium (Approved); nasopharynx / ciliated cells (ciliary rootlets): Medium (Approved)

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 63/100 (63.0%); Glioblastoma Multiforme (validation): 31/58 (53.4%); Kidney Renal Clear Cell Carcinoma (TCGA): 152/521 (29.2%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Force generating protein required for cilia beating in respiratory epithelia (By similarity). Produces force towards the minus ends of microtubules (By similarity). Key component of dynein, a family of motor proteins essential for movement along microtubules (By similarity). Dynein has ATPase activity; the force-producing power stroke is thought to occur on release of ADP (By similarity). Required for structural and functional ... [full text in table]

Curated tissue note. Expressed in airway ciliated epithelial cells (at protein level) (PubMed:31178125, PubMed:33139725). Not detected in spermatozoa (at protein level) (PubMed:31178125)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96DT5 | HPA portal (live; not the pinned values)

DNAH14 | Unsafe / Protein

ENSG00000185842

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); colon / endothelial cells: Low (Uncertain); kidney / cells in tubules: Low (Uncertain); nasopharynx / respiratory epithelial cells: Low (Uncertain)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 78/80 (97.5%); Lung Adenocarcinoma (validation): 95/105 (90.5%); Rectum Adenocarcinoma (validation): 183/207 (88.4%)

Cancer IHC. testis cancer: 1/10 (10.0%); liver cancer: 1/12 (8.3%)

Biology. Force generating protein of respiratory cilia. Produces force towards the minus ends of microtubules. Dynein has ATPase activity; the force-producing power stroke is thought to occur on release of ADP. Involved in sperm motility; implicated in sperm flagellar assembly (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q0VDD8 | HPA portal (live; not the pinned values)

DNAH17 | Unsafe / RNA

ENSG00000187775

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.4 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 33/50 (66.0%); Liver Hepatocellular Carcinoma (validation): 35/231 (15.2%); Head and Neck Squamous Cell Carcinoma (TCGA): 64/492 (13.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Force generating protein component of the outer dynein arms (ODAs) in the sperm flagellum (By similarity). Produces force towards the minus ends of microtubules (By similarity). Key component of dynein, a family of motor proteins essential for movement along microtubules (By similarity). Dynein has ATPase activity; the force-producing power stroke is thought to occur on release of ADP (By similarity). Required for structural and ... [full text in table]

Curated tissue note. Expressed in testis (PubMed:9545504). Expressed in spermatozoa (at protein level). Not detected in airway epithelial cells (at protein level) (PubMed:31178125)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UFH2 | HPA portal (live; not the pinned values)

DNAH2 | Unsafe / Protein

ENSG00000183914

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); nasopharynx / ciliated cells (cilia axoneme): Medium (Enhanced); nasopharynx / ciliated cells (ciliary ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 43/105 (41.0%); Lung Adenocarcinoma (TCGA): 183/497 (36.8%); Colon Adenocarcinoma (validation): 164/486 (33.7%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. As part of the axonemal inner dynein arm complex plays a central role in ciliary beat (PubMed:30811583). Expressed in sperm flagellum, it is required for sperm motility (PubMed:30811583). Dyneins are microtubule-based molecular motors possessing ATPase activities that can convert the chemical energy of ATP into relative sliding between adjacent microtubule doublets to generate ciliary bending (PubMed:30811583)

Curated tissue note. Expressed primarily in trachea and testis, 2 tissues containing axonemal structures. Also expressed in lung. Expressed in spermatozoa (at protein level) (PubMed:31178125)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P225 | HPA portal (live; not the pinned values)

DNAH5 | Unsafe / Protein

ENSG00000039139

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); lung / macrophages: Medium (Enhanced); nasopharynx / ciliated cells (cilia axoneme): High (Enhanced); nasopharynx / ciliated cells (tip of cilia): High (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 35/50 (70.0%); Glioblastoma Multiforme (validation): 21/58 (36.2%); Pancreatic Adenocarcinoma (validation): 25/80 (31.2%)

Cancer IHC. testis cancer: 3/11 (27.3%); pancreatic cancer: 2/9 (22.2%)

Biology. Force generating protein of respiratory cilia (By similarity). Produces force towards the minus ends of microtubules (By similarity). Key component of dynein, a family of motor proteins essential for movement along microtubules (By similarity). Dynein has ATPase activity; the force-producing power stroke is thought to occur on release of ADP (By similarity). Required for structural and functional integrity of the cilia of ependymal ... [full text in table]

Curated tissue note. Expressed in airway epithelial cells (at protein level). Not detected in spermatozoa (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TE73 | HPA portal (live; not the pinned values)

DNAH7 | Unsafe / Protein

ENSG00000118997

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cilia axoneme): Medium (Enhanced); nasopharynx / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 43/58 (74.1%); Breast Invasive Carcinoma (validation): 26/50 (52.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 83/282 (29.4%)

Cancer IHC. lymphoma: 2/12 (16.7%); melanoma: 1/10 (10.0%)

Biology. Force generating protein that plays an important role in respiratory cilia and sperm flagella beating (PubMed:34476482). Produces force towards the minus ends of microtubules. Dynein has ATPase activity; the force-producing power stroke is thought to occur on release of ADP (By similarity)

Curated tissue note. Detected in brain, testis and trachea. Detected in bronchial cells (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WXX0 | HPA portal (live; not the pinned values)

DNAH9 | Unsafe / Protein

ENSG00000007174

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); heart muscle / cardiomyocytes: Low (Enhanced); nasopharynx / ciliated cells (cell body): Low (Enhanced); nasopharynx / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 114/141 (80.9%); Glioblastoma Multiforme (validation): 38/58 (65.5%); Uterine Corpus Endometrial Carcinoma (TCGA): 50/176 (28.4%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Force generating protein required for cilia beating in respiratory epithelia (PubMed:30471717, PubMed:30471718). Produces force towards the minus ends of microtubules (PubMed:30471717, PubMed:30471718). Key component of dynein, a family of motor proteins essential for movement along microtubules (By similarity). Dynein has ATPase activity; the force-producing power stroke is thought to occur on release of ADP (PubMed:30471717, ... [full text in table]

Curated tissue note. Expressed in upper and lower respiratory airway epithelia (at protein level). Not detected in spermatozoa (at protein level) (PubMed:31178125)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NYC9 | HPA portal (live; not the pinned values)

DNAI4 | Unsafe / RNA

ENSG00000152763

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.6 nTPM (limit 2); choroid plexus: 20 nTPM (limit 2); hippocampal formation: 2.6 nTPM (limit 2); hypothalamus: 3 nTPM (limit 2); kidney: 6.1 nTPM (limit 2); lung: 4.1 nTPM (limit 2); parathyroid gland: 6.7 nTPM (limit 6); pituitary gland: 2.7 nTPM (limit 2); retina: 3.9 nTPM (limit ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 53/80 (66.2%); Kidney Renal Papillary Cell Carcinoma (TCGA): 127/282 (45.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays a critical role in the assembly of axonemal dynein complex, thereby playing a role in ciliary motility

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VTH9 | HPA portal (live; not the pinned values)

DNAI7 | Unsafe / Protein

ENSG00000118307

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); lung / macrophages: Medium (Enhanced); nasopharynx / ciliated cells (cell body): Medium (Enhanced); nasopharynx / ciliated cells (tip of cilia): High (Enhanced); thyroid gland ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 39/58 (67.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 47/176 (26.7%); Kidney Renal Papillary Cell Carcinoma (TCGA): 68/282 (24.1%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Via its association with the multisubunit axonemal dynein complex, is potentially involved in the regulation of cilia function. May also act as a cell cycle regulator

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6TDU7 | HPA portal (live; not the pinned values)

DNAJB13 | Unsafe / Protein

ENSG00000187726

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cell body): Medium (Enhanced); nasopharynx / ciliated cells (cilia axoneme): High ... [full text in table]

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 270/349 (77.4%); Uterine Corpus Endometrial Carcinoma (TCGA): 113/176 (64.2%); Ovary Serous Cystadenocarcinoma (validation): 52/81 (64.2%)

Cancer IHC. cervical cancer: 1/9 (11.1%); endometrial cancer: 1/11 (9.1%)

Biology. Functions as part of axonemal radial spoke complexes that play an important part in the motility of sperm and cilia

Curated tissue note. Specifically expressed in testis and trachea

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P59910 | HPA portal (live; not the pinned values)

DNAJC24 | Unsafe / Protein

ENSG00000170946

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / basal cells: Medium (Approved); bronchus / ciliated cells (ciliary rootlets): Medium (Approved); bronchus / ciliated cells (tip of cilia): Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 138/141 (97.9%); Pancreatic Adenocarcinoma (validation): 78/80 (97.5%)

Cancer IHC. melanoma: 7/11 (63.6%); prostate cancer: 4/12 (33.3%)

Biology. Stimulates the ATPase activity of several Hsp70-type chaperones. This ability is enhanced by iron-binding. The iron-bound form is redox-active and can function as electron carrier. Plays a role in the diphthamide biosynthesis, a post-translational modification of histidine which occurs in translation elongation factor 2 (EEF2) which can be ADP-ribosylated by diphtheria toxin and by Pseudomonas exotoxin A (Eta)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6P3W2 | HPA portal (live; not the pinned values)

DNAJC28 | Unsafe / Protein

ENSG00000177692

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: High (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 54/58 (93.1%); Breast Invasive Carcinoma (validation): 41/50 (82.0%); Kidney Chromophobe (TCGA): 2/64 (3.1%)

Cancer IHC. cervical cancer: 10/12 (83.3%); pancreatic cancer: 9/11 (81.8%)

Biology. May have a role in protein folding or as a chaperone

Curated tissue note. Expressed in the fetal and adult brain, testis, uterus, spleen and liver

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NX36 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

DNAJC5B | Unsafe / RNA

ENSG00000147570

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. lung: 5.9 nTPM (limit 2); lymph node: 7.5 nTPM (limit 6); lung: 5.9 nTPM (baseline 2); lymph node: 7.5 nTPM (baseline 6)

Cancer RNA. Lung Adenocarcinoma (validation): 75/105 (71.4%); Lung Squamous Cell Carcinoma (validation): 31/68 (45.6%); Lung Adenocarcinoma (TCGA): 182/497 (36.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UF47 | HPA portal (live; not the pinned values)

DNAL1 | Unsafe / Protein

ENSG00000119661

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: Medium (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / ciliated cells (ciliary rootlets): High (Approved); bronchus / ciliated cells (tip of ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. glioma: 6/9 (66.7%); testis cancer: 6/10 (60.0%)

Biology. Part of the multisubunit axonemal ATPase complexes that generate the force for cilia motility and govern beat frequency (By similarity). Component of the outer arm dynein (ODA). May be involved in a mechanosensory feedback mechanism controlling ODA activity based on external conformational cues by tethering the outer arm dynein heavy chain (DNAH5) to the microtubule within the axoneme (By similarity). Important for ciliary function in ... [full text in table]

Curated tissue note. Expressed in tissues carrying motile cilia such as respiratory epithelia, ependyma and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4LDG9 | HPA portal (live; not the pinned values)

DNALI1 | Unsafe / Protein

ENSG00000163879

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Low (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); colon / enterocytes: Low (Enhanced); duodenum / enterocytes: Medium (Enhanced); duodenum / enterocytes - Gradient: Ascending ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 138/141 (97.9%); Breast Invasive Carcinoma (validation): 47/50 (94.0%)

Cancer IHC. endometrial cancer: 1/12 (8.3%); ovarian cancer: 1/12 (8.3%)

Biology. Involved in sperm flagellum assembly

Curated tissue note. Expressed in many tissues. A smaller 0.9 kb and a larger 2.5 kb transcripts were detected at the highest level in the testis, at medium levels in the prostate, heart, liver, lung and pancreas, at low levels in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14645 | HPA portal (live; not the pinned values)

DNASE2B | Unsafe / RNA

ENSG00000137976

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. lung: 2.3 nTPM (limit 2); salivary gland: 165.3 nTPM (limit 6); lung: 2.3 nTPM (baseline 2); salivary gland: 165.3 nTPM (baseline 6)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 360/480 (75.0%); Skin Cuteneous Melanoma (TCGA): 18/99 (18.2%); Breast Invasive Carcinoma (validation): 4/50 (8.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Hydrolyzes DNA under acidic conditions. Does not require divalent cations for activity. Participates in the degradation of nuclear DNA during lens cell differentiation

Curated tissue note. Highly expressed in the eye lens and in salivary gland. Detected at lower levels in lung, prostate and lymph node. Isoform 2 is lung specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WZ79 | HPA portal (live; not the pinned values)

DND1 | Unsafe / RNA

ENSG00000256453

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 3 nTPM (limit 2); cerebellum: 3 nTPM (baseline 2)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 216/231 (93.5%); Breast Invasive Carcinoma (validation): 44/50 (88.0%); Prostate Adenocarcinoma (TCGA): 389/480 (81.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. RNA-binding factor that positively regulates gene expression by prohibiting miRNA-mediated gene suppression. Relieves miRNA repression in germline cells (By similarity). Prohibits the function of several miRNAs by blocking the accessibility of target mRNAs. Sequence-specific RNA-binding factor that binds specifically to U-rich regions (URRs) in the 3' untranslated region (3'-UTR) of several mRNAs. Does not bind to miRNAs. May play a ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYX4 | HPA portal (live; not the pinned values)

DOCK1 | Unsafe / Protein

ENSG00000150760

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. glioma: 9/11 (81.8%); skin cancer: 8/11 (72.7%)

Biology. Involved in cytoskeletal rearrangements required for phagocytosis of apoptotic cells and cell motility. Along with DOCK1, mediates CRK/CRKL regulation of epithelial and endothelial cell spreading and migration on type IV collagen (PubMed:19004829). Functions as a guanine nucleotide exchange factor (GEF), which activates Rac Rho small GTPases by exchanging bound GDP for free GTP. Its GEF activity may be enhanced by ELMO1 (PubMed:8657152)

Curated tissue note. Highly expressed in placenta, lung, kidney, pancreas and ovary. Expressed at intermediate level in thymus, testes and colon

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14185 | HPA portal (live; not the pinned values)

DOK6 | Unsafe / Protein

ENSG00000206052

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); cerebral cortex / neuropil: Medium (Approved); colon / endothelial cells: Low (Approved); colon / peripheral nerve/ganglion: Low (Approved); heart muscle / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 32/58 (55.2%); Kidney Renal Clear Cell Carcinoma (validation): 23/100 (23.0%); Lung Squamous Cell Carcinoma (validation): 8/68 (11.8%)

Cancer IHC. lymphoma: 4/11 (36.4%); liver cancer: 1/10 (10.0%)

Biology. DOK proteins are enzymatically inert adaptor or scaffolding proteins. They provide a docking platform for the assembly of multimolecular signaling complexes. DOK6 promotes Ret-mediated neurite growth. May have a role in brain development and/or maintenance

Curated tissue note. Highly expressed in fetal and adult brain. Highly expressed in the cerebellum. Weak expression in kidney, spinal cord and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PKX4 | HPA portal (live; not the pinned values)

DPH6 | Unsafe / Protein

ENSG00000134146

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Approved); adrenal gland / glandular cells: High (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebral cortex / glial cells: Low (Approved); colon / endothelial cells: Low (Approved); colon / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 140/141 (99.3%); Colon Adenocarcinoma (validation): 479/486 (98.6%); Lung Adenocarcinoma (validation): 103/105 (98.1%)

Cancer IHC. liver cancer: 9/12 (75.0%); testis cancer: 5/10 (50.0%)

Biology. Amidase that may catalyze the last step of diphthamide biosynthesis using ammonium and ATP (PubMed:23169644). Diphthamide biosynthesis consists in the conversion of an L-histidine residue in the translation elongation factor (EEF2) to diphthamide (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7L8W6 | HPA portal (live; not the pinned values)

DPPA4 | Unsafe / RNA

ENSG00000121570

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. thymus: 12.1 nTPM (limit 10)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 121/133 (91.0%); Pancreatic Adenocarcinoma (validation): 6/80 (7.5%); Glioblastoma Multiforme (validation): 1/58 (1.7%)

Cancer IHC. testis cancer: 12/12 (100.0%); urothelial cancer: 4/12 (33.3%)

Biology. May be involved in the maintenance of active epigenetic status of target genes. May inhibit differentiation of embryonic cells into a primitive ectoderm lineage

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7L190 | HPA portal (live; not the pinned values)

DRC7 | Unsafe / RNA

ENSG00000159625

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.3 nTPM (limit 2); choroid plexus: 12 nTPM (limit 2); hypothalamus: 2.1 nTPM (limit 2); basal ganglia: 2.3 nTPM (baseline 2); choroid plexus: 12 nTPM (baseline 2); hypothalamus: 2.1 nTPM (baseline 2)

Cancer RNA. Kidney Renal Papillary Cell Carcinoma (TCGA): 175/282 (62.1%); Uterine Corpus Endometrial Carcinoma (TCGA): 51/176 (29.0%); Ovary Serous Cystadenocarcinoma (TCGA): 36/349 (10.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of the nexin-dynein regulatory complex (N-DRC) a key regulator of ciliary/flagellar motility which maintains the alignment and integrity of the distal axoneme and regulates microtubule sliding in motile axonemes (By similarity). Involved in the regulation of flagellar motility (By similarity). Essential for male fertility, sperm head morphogenesis and sperm flagellum formation (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IY82 | HPA portal (live; not the pinned values)

DRD3 | Unsafe / RNA

ENSG00000151577

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 3.2 nTPM (limit 2); basal ganglia: 3.2 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. G protein-coupled receptor for dopamine, a catecholamine neurotransmitter hormone that functions as the brain's 'reward chemical', driving motivation, reinforcement learning and pleasure (PubMed:19520868, PubMed:33548201, PubMed:37221270, PubMed:39237617, PubMed:39984436, PubMed:8301582). Dopamine binding causes a conformation change that triggers signaling via guanine nucleotide-binding proteins (G proteins) and modulates the activity ... [full text in table]

Curated tissue note. Brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P35462 | HPA portal (live; not the pinned values)

DRICH1 | Unsafe / RNA

ENSG00000189269

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 3.7 nTPM (limit 2); kidney: 2.5 nTPM (limit 2); pancreas: 2.3 nTPM (limit 2); parathyroid gland: 8.9 nTPM (limit 6); retina: 3.8 nTPM (limit 2); bone marrow: 3.7 nTPM (baseline 2); kidney: 2.5 nTPM (baseline 2); pancreas: 2.3 nTPM (baseline 2); parathyroid gland: 8.9 nTPM (baseline 6); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 8/58 (13.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 35/282 (12.4%); Testicular Germ Cell Tumor (TCGA): 14/133 (10.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PGQ1 | HPA portal (live; not the pinned values)

DTL | Unsafe / Protein

ENSG00000143476

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: High (Supported); esophagus / squamous epithelial cells: Low (Supported); lung / macrophages: Low (Supported); lymph node / germinal center cells: Medium (Supported); lymph node / non-germinal center cells: Low (Supported); skin 2 / epidermal cells: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 479/486 (98.6%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Lung Squamous Cell Carcinoma (validation): 65/68 (95.6%)

Cancer IHC. colorectal cancer: 2/12 (16.7%); melanoma: 1/11 (9.1%)

Biology. Substrate-specific adapter of a DCX (DDB1-CUL4-X-box) E3 ubiquitin-protein ligase complex required for cell cycle control, DNA damage response and translesion DNA synthesis. The DCX(DTL) complex, also named CRL4(CDT2) complex, mediates the polyubiquitination and subsequent degradation of CDT1, CDKN1A/p21(CIP1), FBH1, KMT5A and SDE2 (PubMed:16861906, PubMed:16949367, PubMed:16964240, PubMed:17085480, PubMed:18703516, PubMed:18794347, ... [full text in table]

Curated tissue note. Expressed in placenta and testis, very low expression seen in skeletal muscle. Detected in all hematopoietic tissues examined, with highest expression in thymus and bone marrow. A low level detected in the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NZJ0 | HPA portal (live; not the pinned values)

DTWD2 | Unsafe / Protein

ENSG00000169570

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / ciliated cells (cell body): Medium (Approved); colon / glandular cells: High (Approved); duodenum / glandular cells: Medium (Approved); esophagus / squamous epithelial cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Pancreatic Adenocarcinoma (validation): 68/80 (85.0%)

Cancer IHC. pancreatic cancer: 7/10 (70.0%); breast cancer: 7/12 (58.3%)

Biology. Catalyzes the formation of 3-(3-amino-3-carboxypropyl)uridine (acp3U) at position 20a in the D-loop of several cytoplasmic tRNAs (acp3U(20a)) (PubMed:31804502, PubMed:39173631). Also has a weak activity to form acp3U at position 20 in the D-loop of tRNAs (acp3U(20)) (PubMed:31804502). Involved in glycoRNA biosynthesis by mediating formation of acp3U, which acts as an attachment site for N-glycans on tRNAs (PubMed:39173631). GlycoRNAs ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NBA8 | HPA portal (live; not the pinned values)

DUSP19 | Unsafe / Protein

ENSG00000162999

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebral cortex / neuronal cells: Low (Approved); colon / glandular cells: High (Approved); colon / peripheral nerve/ganglion: Low (Approved); duodenum / glandular cells: High (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 14/50 (28.0%); Pancreatic Adenocarcinoma (validation): 10/80 (12.5%)

Cancer IHC. renal cancer: 5/12 (41.7%); colorectal cancer: 4/11 (36.4%)

Biology. Has a dual specificity toward Ser/Thr and Tyr-containing proteins

Curated tissue note. Expressed in the heart, lung, liver, and pancreas. The expression level in the pancreas is the highest

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WTR2 | HPA portal (live; not the pinned values)

DYNLRB2 | Unsafe / RNA

ENSG00000168589

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.4 nTPM (limit 2); amygdala: 8.8 nTPM (limit 2); basal ganglia: 11.3 nTPM (limit 2); cerebral cortex: 3.3 nTPM (limit 2); choroid plexus: 56.3 nTPM (limit 2); hippocampal formation: 11.5 nTPM (limit 2); hypothalamus: 11.4 nTPM (limit 2); kidney: 5.6 nTPM (limit 2); lung: 5.9 nTPM ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 45/50 (90.0%); Breast Invasive Carcinoma (TCGA): 466/1022 (45.6%); Kidney Renal Papillary Cell Carcinoma (TCGA): 112/282 (39.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Acts as one of several non-catalytic accessory components of the cytoplasmic dynein 1 complex that are thought to be involved in linking dynein to cargos and to adapter proteins that regulate dynein function. Cytoplasmic dynein 1 acts as a motor for the intracellular retrograde motility of vesicles and organelles along microtubules

Curated tissue note. High expression in heart, brain, placenta, skeletal muscle, prostate and small intestine; moderate in kidney, pancreas, spleen, testis, ovary and colon; low in lung, liver, thymus and leukocyte

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TF09 | HPA portal (live; not the pinned values)

DYNLT2 | Unsafe / High RNA

ENSG00000184786

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 18.3 nTPM; fallopian tube/t-cells; hpa_single_cell_tissue; vital; 41.52 TPM; blood/CD4_STIM; dice_cd4_stim; vital

Cancer RNA. Glioblastoma Multiforme (validation): 2/58 (3.4%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of dynein, a family of motor proteins essential for movement along microtubules (PubMed:11278908, PubMed:12584439). Required for structural and functional integrity of cilia (By similarity). Candidate for involvement in male sterility (By similarity)

Curated tissue note. Expressed predominantly in testis. Also expressed in brain, lung and trachea

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZS6 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

DYRK3 | Unsafe / RNA

ENSG00000143479

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.3 nTPM (limit 2); bone marrow: 8.7 nTPM (limit 2); cerebral cortex: 2.5 nTPM (limit 2); choroid plexus: 8.1 nTPM (limit 2); heart muscle: 8 nTPM (limit 2); hippocampal formation: 2.7 nTPM (limit 2); hypothalamus: 2.5 nTPM (limit 2); kidney: 3.2 nTPM (limit 2); liver: 4 nTPM (limit ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 50/50 (100.0%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Pancreatic Adenocarcinoma (validation): 75/80 (93.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Dual-specificity protein kinase that promotes disassembly of several types of membraneless organelles during mitosis, such as stress granules, nuclear speckles and pericentriolar material (PubMed:29973724). Dual-specificity tyrosine-regulated kinases (DYRKs) autophosphorylate a critical tyrosine residue in their activation loop and phosphorylate their substrate on serine and threonine residues (PubMed:29634919, PubMed:9748265). Acts as ... [full text in table]

Curated tissue note. Isoform 1: Highly expressed in testis and in hematopoietic tissue such as fetal liver, and bone marrow (PubMed:10779429). Isoform 1: Predominant form in fetal liver and bone marrow (PubMed:10779429). Isoform ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43781 | HPA portal (live; not the pinned values)

DZIP3 | Unsafe / Protein

ENSG00000198919

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / ciliated cells (cell body): Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Low (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 203/207 (98.1%); Colon Adenocarcinoma (validation): 473/486 (97.3%)

Cancer IHC. melanoma: 5/9 (55.6%); ovarian cancer: 4/12 (33.3%)

Biology. E3 Ubiquitin ligase proteins mediate ubiquitination and subsequent proteasomal degradation of target proteins. E3 ubiquitin ligases accept ubiquitin from an E2 ubiquitin-conjugating enzyme in the form of a thioester and then directly transfers the ubiquitin to targeted substrates. Able to specifically bind RNA

Curated tissue note. Widely expressed at low level. Highly expressed in skeletal muscle, kidney and heart. Expressed at low level in placenta, lung, brain, liver and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86Y13 | HPA portal (live; not the pinned values)

E2F7 | Unsafe / RNA

ENSG00000165891

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 4.9 nTPM (limit 2); bone marrow: 4.9 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 50/58 (86.2%); Colon Adenocarcinoma (validation): 414/486 (85.2%); Rectum Adenocarcinoma (validation): 155/207 (74.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Atypical E2F transcription factor that participates in various processes such as angiogenesis, polyploidization of specialized cells and DNA damage response. Mainly acts as a transcription repressor that binds DNA independently of DP proteins and specifically recognizes the E2 recognition site 5'-TTTC[CG]CGC-3'. Directly represses transcription of classical E2F transcription factors such as E2F1. Acts as a regulator of S-phase by ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96AV8 | HPA portal (live; not the pinned values)

EBI3 | Unsafe / RNA

ENSG00000105246

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.8 nTPM (limit 2); choroid plexus: 4.4 nTPM (limit 2); hippocampal formation: 2.5 nTPM (limit 2); hypothalamus: 2.2 nTPM (limit 2); midbrain: 3.8 nTPM (limit 2); spinal cord: 5 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 47/50 (94.0%); Kidney Renal Clear Cell Carcinoma (validation): 79/100 (79.0%); Glioblastoma Multiforme (TCGA): 108/141 (76.6%)

Cancer IHC. skin cancer: 1/10 (10.0%); stomach cancer: 1/11 (9.1%)

Biology. Associates with IL27 to form the IL-27 interleukin, a heterodimeric cytokine which functions in innate immunity. IL-27 has pro- and anti-inflammatory properties, that can regulate T-helper cell development, suppress T-cell proliferation, stimulate cytotoxic T-cell activity, induce isotype switching in B-cells, and that has diverse effects on innate immune cells. Among its target cells are CD4 T-helper cells which can differentiate in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14213 | HPA portal (live; not the pinned values)

EDAR | Unsafe / Protein

ENSG00000135960

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 116/207 (56.0%); Colon Adenocarcinoma (validation): 249/486 (51.2%); Rectum Adenocarcinoma (TCGA): 38/88 (43.2%)

Cancer IHC. colorectal cancer: 10/12 (83.3%); stomach cancer: 10/12 (83.3%)

Biology. Receptor for EDA isoform A1, but not for EDA isoform A2. Mediates the activation of NF-kappa-B and JNK. May promote caspase-independent cell death

Curated tissue note. Detected in fetal kidney, lung, skin and cultured neonatal epidermal keratinocytes. Not detected in lymphoblast and fibroblast cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UNE0 | HPA portal (live; not the pinned values)

EDDM3A | Unsafe / High RNA

ENSG00000181562

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 31.1 nTPM; unsited/Collecting duct cells; hpa_single_cell; unresolved; 5.182 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 3/50 (6.0%); Kidney Chromophobe (TCGA): 2/64 (3.1%); Liver Hepatocellular Carcinoma (TCGA): 6/362 (1.7%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Possible function in sperm maturation

Curated tissue note. Epididymis, with predominant expression in the corpus region. Moderately expressed in the vas deferens; only low levels are detectable in the caput and cauda regions

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14507 | HPA portal (live; not the pinned values)

EFCAB10 | Unsafe / RNA

ENSG00000185055

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 3.4 nTPM (limit 2); bone marrow: 2.3 nTPM (limit 2); choroid plexus: 12.4 nTPM (limit 2); hippocampal formation: 2.7 nTPM (limit 2); hypothalamus: 3.3 nTPM (limit 2); lung: 3.1 nTPM (limit 2); pituitary gland: 2.1 nTPM (limit 2); basal ganglia: 3.4 nTPM (baseline 2); bone marrow: 2.3 ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 342/480 (71.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 117/176 (66.5%); Liver Hepatocellular Carcinoma (validation): 151/231 (65.4%)

Cancer IHC. melanoma: 12/12 (100.0%); renal cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NFE3 | HPA portal (live; not the pinned values)

EFCAB11 | Unsafe / RNA

ENSG00000140025

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4 nTPM (limit 2); amygdala: 7.6 nTPM (limit 2); basal ganglia: 6.3 nTPM (limit 2); bone marrow: 5.9 nTPM (limit 2); cerebellum: 2.1 nTPM (limit 2); cerebral cortex: 4.9 nTPM (limit 2); choroid plexus: 5.5 nTPM (limit 2); duodenum: 4.9 nTPM (limit 2); heart muscle: 2.5 nTPM (limit 2); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (TCGA): 87/88 (98.9%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 276/283 (97.5%); Breast Invasive Carcinoma (TCGA): 980/1022 (95.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BUY7 | HPA portal (live; not the pinned values)

EFCAB12 | Unsafe / RNA

ENSG00000172771

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.8 nTPM (limit 2); choroid plexus: 10.5 nTPM (limit 2); hypothalamus: 3 nTPM (limit 2); pituitary gland: 3.5 nTPM (limit 2); basal ganglia: 2.8 nTPM (baseline 2); choroid plexus: 10.5 nTPM (baseline 2); hypothalamus: 3 nTPM (baseline 2); pituitary gland: 3.5 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 44/176 (25.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 55/282 (19.5%); Prostate Adenocarcinoma (TCGA): 47/480 (9.8%)

Cancer IHC. colorectal cancer: 10/10 (100.0%); endometrial cancer: 10/11 (90.9%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6NXP0 | HPA portal (live; not the pinned values)

EFCAB13 | Unsafe / RNA

ENSG00000178852

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.5 nTPM (limit 2); bone marrow: 3.8 nTPM (limit 2); duodenum: 4.6 nTPM (limit 2); heart muscle: 2.4 nTPM (limit 2); kidney: 5.2 nTPM (limit 2); liver: 3.3 nTPM (limit 2); lung: 3 nTPM (limit 2); pituitary gland: 2.5 nTPM (limit 2); retina: 3.8 nTPM (limit 2); small intestine: 3.3 ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 164/207 (79.2%); Thyroid Carcinoma (TCGA): 366/495 (73.9%); Colon Adenocarcinoma (validation): 346/486 (71.2%)

Cancer IHC. pancreatic cancer: 8/10 (80.0%); testis cancer: 6/11 (54.5%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IY85 | HPA portal (live; not the pinned values)

EFCAB2 | Unsafe / Protein

ENSG00000203666

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: High (Approved); cerebellum / cells in granular layer: Medium (Approved); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 67/68 (98.5%); Glioblastoma Multiforme (TCGA): 137/141 (97.2%)

Cancer IHC. endometrial cancer: 11/12 (91.7%); colorectal cancer: 10/12 (83.3%)

Biology. Component of the nexin-dynein regulatory complex (N-DRC), a key regulator of ciliary/flagellar motility which maintains the alignment and integrity of the distal axoneme and regulates microtubule sliding in motile axonemes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VUJ9 | HPA portal (live; not the pinned values)

EFCAB5 | Unsafe / High RNA

ENSG00000176927

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 34.1 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 6.841 TPM; blood/eosinophil; hpa_immune_cell_samples; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 3/50 (6.0%); Rectum Adenocarcinoma (validation): 10/207 (4.8%); Colon Adenocarcinoma (validation): 19/486 (3.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A4FU69 | HPA portal (live; not the pinned values)

EFCAB6 | Unsafe / RNA

ENSG00000186976

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3.1 nTPM (limit 2); retina: 3 nTPM (limit 2); choroid plexus: 3.1 nTPM (baseline 2); retina: 3 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 30/50 (60.0%); Glioblastoma Multiforme (validation): 28/58 (48.3%); Pancreatic Adenocarcinoma (validation): 8/80 (10.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Negatively regulates the androgen receptor by recruiting histone deacetylase complex, and protein DJ-1 antagonizes this inhibition by abrogation of this complex (PubMed:12612053). Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia axoneme, which is required for motile cilia beating (By similarity)

Curated tissue note. Specifically expressed in the testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5THR3 | HPA portal (live; not the pinned values)

EGFL6 | Unsafe / RNA

ENSG00000198759

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. lung: 24.8 nTPM (limit 12); pituitary gland: 2.5 nTPM (limit 2); retina: 2.2 nTPM (limit 2)

Cancer RNA. Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 67/68 (98.5%); Lung Squamous Cell Carcinoma (TCGA): 441/489 (90.2%)

Cancer IHC. prostate cancer: 3/11 (27.3%); stomach cancer: 1/11 (9.1%)

Biology. May bind integrin alpha-8/beta-1 and play a role in hair follicle morphogenesis. Promotes matrix assembly (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IUX8 | HPA portal (live; not the pinned values)

EGR4 | Unsafe / Protein

ENSG00000135625

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: Low (Enhanced); cerebellum / cells in granular layer: Low (Enhanced); cerebral cortex / glial cells: Low (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); hippocampus / glial cells: Low (Enhanced)

Cancer RNA. Stomach Adenocarcinoma (TCGA): 13/346 (3.8%); Breast Invasive Carcinoma (validation): 1/50 (2.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 3/176 (1.7%)

Cancer IHC. melanoma: 2/10 (20.0%); glioma: 1/10 (10.0%)

Biology. Transcriptional regulator. Recognizes and binds to the DNA sequence 5'-GCGGGGGCG-3' (GSG). Activates the transcription of target genes whose products are required for mitogenesis and differentiation (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q05215 | HPA portal (live; not the pinned values)

EID2B | Unsafe / Protein

ENSG00000176401

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); cerebral cortex / endothelial cells: Low (Approved); colon / endothelial cells: Medium (Approved); colon / glandular cells: Medium (Approved); duodenum / glandular cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 39/50 (78.0%); Glioblastoma Multiforme (TCGA): 84/141 (59.6%)

Cancer IHC. urothelial cancer: 10/12 (83.3%); skin cancer: 8/10 (80.0%)

Biology. Acts as a repressor of MYOD-dependent transcription, glucocorticoid receptor-dependent transcription, and muscle differentiation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96D98 | HPA portal (live; not the pinned values)

EID3 | Unsafe / Protein

ENSG00000255150

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebellum / Purkinje cells: Low (Uncertain); colon / glandular cells: Low (Uncertain); heart muscle / cardiomyocytes: Low (Uncertain); kidney / cells in tubules: Low (Uncertain); rectum / glandular cells: Low (Uncertain); skin 1 / keratinocytes: Medium (Uncertain); skin 2 / epidermal cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 54/58 (93.1%); Breast Invasive Carcinoma (validation): 40/50 (80.0%); Testicular Germ Cell Tumor (TCGA): 78/133 (58.6%)

Cancer IHC. melanoma: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. Tissue-specific component of the SMC5-SMC6 complex, a complex involved in repair of DNA double-strand breaks by homologous recombination. The complex may promote sister chromatid homologous recombination by recruiting the SMC1-SMC3 cohesin complex to double-strand breaks. The complex is required for telomere maintenance via recombination and mediates sumoylation of shelterin complex (telosome) components Acts as a repressor of nuclear ... [full text in table]

Curated tissue note. Highly expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N140 | HPA portal (live; not the pinned values)

EIF4E | Unsafe / Protein

ENSG00000151247

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Supported); adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: Medium (Supported); bronchus / respiratory epithelial cells: Medium (Supported); caudate / glial cells: Low (Supported); caudate / neuronal cells: Low (Supported); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); glioma: 12/12 (100.0%)

Biology. Acts in the cytoplasm to initiate and regulate protein synthesis and is required in the nucleus for export of a subset of mRNAs from the nucleus to the cytoplasm which promotes processes such as RNA capping, processing and splicing (PubMed:11606200, PubMed:22578813, PubMed:22684010, PubMed:24335285, PubMed:29987188). Component of the protein complex eIF4F, which is involved in the recognition of the mRNA cap, ATP-dependent unwinding of ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P06730 | HPA portal (live; not the pinned values)

ELAVL2 | Unsafe / Protein

ENSG00000107105

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Medium (Enhanced); cerebellum / GLUC cells - cytoplasm/membrane: High (Enhanced); cerebellum / GLUC cells - nucleus: High (Enhanced); cerebellum / granular cells - cytoplasm/membrane: Low (Enhanced); cerebellum / molecular layer cells - cytoplasm/membrane: Low (Enhanced); ... [full text in table]

Cancer RNA. Kidney Chromophobe (TCGA): 41/64 (64.1%); Glioblastoma Multiforme (validation): 36/58 (62.1%); Testicular Germ Cell Tumor (TCGA): 75/133 (56.4%)

Cancer IHC. colorectal cancer: 3/12 (25.0%); urothelial cancer: 3/12 (25.0%)

Biology. RNA-binding protein that binds to the 3' untranslated region (3'UTR) of target mRNAs (By similarity). Seems to recognize a GAAA motif (By similarity). Can bind to its own 3'UTR, the FOS 3'UTR and the ID 3'UTR (By similarity)

Curated tissue note. Brain; neural-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q12926 | HPA portal (live; not the pinned values)

ELL3 | Unsafe / Protein

ENSG00000128886

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): High (Approved); bronchus / ciliated cells (ciliary rootlets): High (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / neuronal cells: Low (Approved); duodenum / glandular cells: Medium (Approved); hippocampus / neuronal cells: ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 48/50 (96.0%); Prostate Adenocarcinoma (TCGA): 421/480 (87.7%); Pancreatic Adenocarcinoma (validation): 48/80 (60.0%)

Cancer IHC. colorectal cancer: 8/12 (66.7%); ovarian cancer: 5/10 (50.0%)

Biology. Enhancer-binding elongation factor that specifically binds enhancers in embryonic stem cells (ES cells), marks them, and is required for their future activation during stem cell specification. Does not only bind to enhancer regions of active genes, but also marks the enhancers that are in a poised or inactive state in ES cells and is required for establishing proper RNA polymerase II occupancy at developmentally regulated genes in a ... [full text in table]

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HB65 | HPA portal (live; not the pinned values)

ELOA2 | Unsafe / Protein

ENSG00000206181

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / neuronal cells: Low (Approved); colon / glandular cells: Medium (Approved); duodenum / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. liver cancer: 5/12 (41.7%); melanoma: 4/10 (40.0%)

Biology. SIII, also known as elongin, is a general transcription elongation factor that increases the RNA polymerase II transcription elongation past template-encoded arresting sites. Subunit A2 is transcriptionally active but its transcription activity is not enhanced by binding to the dimeric complex of the SIII regulatory subunits B and C (elongin BC complex)

Curated tissue note. Specifically expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYF1 | HPA portal (live; not the pinned values)

ELOVL2 | Unsafe / RNA

ENSG00000197977

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 10.9 nTPM (limit 2); basal ganglia: 12.6 nTPM (limit 2); cerebellum: 5.8 nTPM (limit 2); cerebral cortex: 13.1 nTPM (limit 2); heart muscle: 3.1 nTPM (limit 2); hippocampal formation: 7.1 nTPM (limit 2); hypothalamus: 5.3 nTPM (limit 2); liver: 50.9 nTPM (limit 2); midbrain: 7.8 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 53/58 (91.4%); Liver Hepatocellular Carcinoma (validation): 209/231 (90.5%); Glioblastoma Multiforme (TCGA): 123/141 (87.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Catalyzes the first and rate-limiting reaction of the four reactions that constitute the long-chain fatty acids elongation cycle. This endoplasmic reticulum-bound enzymatic process allows the addition of 2 carbons to the chain of long- and very long-chain fatty acids (VLCFAs) per cycle. Condensing enzyme that catalyzes the synthesis of polyunsaturated very long chain fatty acid (C20- and C22-PUFA), acting specifically toward ... [full text in table]

Curated tissue note. Liver and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NXB9 | HPA portal (live; not the pinned values)

ELSPBP1 | Unsafe / RNA

ENSG00000169393

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 72.3 nTPM (limit 2)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 7/99 (7.1%); Kidney Renal Papillary Cell Carcinoma (TCGA): 19/282 (6.7%); Kidney Chromophobe (TCGA): 4/64 (6.2%)

Cancer IHC. testis cancer: 1/12 (8.3%); breast cancer: 0/10 (0.0%)

Biology. Binds to spermatozoa upon ejaculation and may play a role in sperm capacitation. Has phosphorylcholine-binding activity (By similarity)

Curated tissue note. Detected in cauda epididymidal fluid and on sperm membrane (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96BH3 | HPA portal (live; not the pinned values)

EME1 | Unsafe / Protein

ENSG00000154920

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / glial cells: High (Supported); cerebellum / cells in granular layer: High ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 44/50 (88.0%); Liver Hepatocellular Carcinoma (validation): 201/231 (87.0%); Testicular Germ Cell Tumor (TCGA): 99/133 (74.4%)

Cancer IHC. breast cancer: 12/12 (100.0%); lymphoma: 12/12 (100.0%)

Biology. Non-catalytic subunit of the structure-specific, heterodimeric DNA endonuclease MUS81-EME1 which is involved in the maintenance of genome stability. In the complex, EME1 is required for DNA cleavage, participating in DNA recognition and bending (PubMed:12686547, PubMed:12721304, PubMed:14617801, PubMed:17289582, PubMed:24733841, PubMed:24813886, PubMed:35290797, PubMed:39015284). MUS81-EME1 cleaves 3'-flaps and nicked Holliday ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96AY2 | HPA portal (live; not the pinned values)

ENAM | Unsafe / RNA

ENSG00000132464

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. heart muscle: 6.3 nTPM (limit 2); kidney: 8.6 nTPM (limit 2); pancreas: 3.5 nTPM (limit 2); skeletal muscle: 4.7 nTPM (limit 2); heart muscle: 6.3 nTPM (baseline 2); kidney: 8.6 nTPM (baseline 2); pancreas: 3.5 nTPM (baseline 2); skeletal muscle: 4.7 nTPM (baseline 2)

Cancer RNA. Kidney Chromophobe (TCGA): 35/64 (54.7%); Kidney Renal Papillary Cell Carcinoma (TCGA): 93/282 (33.0%); Kidney Renal Clear Cell Carcinoma (validation): 23/100 (23.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Involved in the mineralization and structural organization of enamel. Involved in the extension of enamel during the secretory stage of dental enamel formation

Curated tissue note. Expressed in tooth particularly in odontoblast, ameloblast and cementoblast

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NRM1 | HPA portal (live; not the pinned values)

ENDOV | Unsafe / RNA

ENSG00000173818

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 8.1 nTPM (limit 2); amygdala: 10.2 nTPM (limit 2); basal ganglia: 12.9 nTPM (limit 2); bone marrow: 4.9 nTPM (limit 2); cerebellum: 11.4 nTPM (limit 2); cerebral cortex: 12.1 nTPM (limit 2); choroid plexus: 8.8 nTPM (limit 2); colon: 10.6 nTPM (limit 6); duodenum: 6.1 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%); Prostate Adenocarcinoma (TCGA): 475/480 (99.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Endoribonuclease that specifically cleaves inosine-containing RNAs: cleaves RNA at the second phosphodiester bond 3' to inosine (PubMed:23912683, PubMed:23912718, PubMed:25195743, PubMed:27573237, PubMed:31703097). Active against both single-stranded and double-stranded RNAs (PubMed:25195743, PubMed:31703097). Has strong preference for single-stranded RNAs (ssRNAs) toward double-stranded RNAs (dsRNAs) (PubMed:23912718). Cleaves mRNAs ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N8Q3 | HPA portal (live; not the pinned values)

ENPP5 | Unsafe / Protein

ENSG00000112796

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / cells in granular layer: Low (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 492/495 (99.4%); Prostate Adenocarcinoma (TCGA): 472/480 (98.3%); Lung Adenocarcinoma (validation): 102/105 (97.1%)

Cancer IHC. prostate cancer: 12/12 (100.0%); testis cancer: 11/11 (100.0%)

Biology. Can hydrolyze NAD but cannot hydrolyze nucleotide di- and triphosphates. Lacks lysopholipase D activity. May play a role in neuronal cell communication

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UJA9 | HPA portal (live; not the pinned values)

ENTPD4 | Unsafe / Protein

ENSG00000197217

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Supported); bronchus / basal cells: Medium (Supported); bronchus / ciliated cells (cell body): High (Supported); caudate / neuronal cells: Medium (Supported); cerebellum / cells in molecular layer: Medium (Supported); cerebellum / Purkinje cells: High ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. endometrial cancer: 12/12 (100.0%); prostate cancer: 12/12 (100.0%)

Biology. Catalyzes the hydrolysis of nucleoside triphosphates and diphosphates in a calcium- or magnesium-dependent manner, with a preference for pyrimidines. Preferentially hydrolyzes UTP and TTP. AMP, ADP, ATP and UMP are not substrates (PubMed:10858452, PubMed:9556635). Preferentially activated by Ca(2+) over Mg(2+) (PubMed:10858452) Has a broad substrate specificity with the ability of cleaving all nucleotide di- and triphosphates with the ... [full text in table]

Curated tissue note. Ubiquitous. Highest expression in testis and lowest in bladder

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y227 | HPA portal (live; not the pinned values)

EPC1 | Unsafe / RNA

ENSG00000120616

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 13.9 nTPM (limit 10); adrenal gland: 9.3 nTPM (limit 2); amygdala: 7.9 nTPM (limit 2); basal ganglia: 8.5 nTPM (limit 2); bone marrow: 147.7 nTPM (limit 2); cerebellum: 21.4 nTPM (limit 2); cerebral cortex: 9.6 nTPM (limit 2); choroid plexus: 9.9 nTPM (limit 2); colon: 15.4 nTPM ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of the NuA4 histone acetyltransferase (HAT) complex, a multiprotein complex involved in transcriptional activation of select genes principally by acetylation of nucleosomal histones H4 and H2A (PubMed:14966270). The NuA4 complex plays a direct role in repair of DNA double-strand breaks (DSBs) by promoting homologous recombination (HR) (PubMed:27153538). The NuA4 complex is also required for spermatid development by promoting ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H2F5 | HPA portal (live; not the pinned values)

EPHA6 | Unsafe / RNA

ENSG00000080224

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. cerebral cortex: 2.1 nTPM (limit 2); cerebral cortex: 2.1 nTPM (baseline 2)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 86/480 (17.9%); Kidney Renal Clear Cell Carcinoma (validation): 15/100 (15.0%); Glioblastoma Multiforme (validation): 8/58 (13.8%)

Cancer IHC. colorectal cancer: 9/11 (81.8%); endometrial cancer: 7/11 (63.6%)

Biology. Receptor tyrosine kinase which binds promiscuously GPI-anchored ephrin-A family ligands residing on adjacent cells, leading to contact-dependent bidirectional signaling into neighboring cells. The signaling pathway downstream of the receptor is referred to as forward signaling while the signaling pathway downstream of the ephrin ligand is referred to as reverse signaling (By similarity)

Curated tissue note. Expressed in brain and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UF33 | HPA portal (live; not the pinned values)

EPHA8 | Unsafe / RNA

ENSG00000070886

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.4 nTPM (limit 2); choroid plexus: 32.4 nTPM (limit 2); hypothalamus: 2.7 nTPM (limit 2); midbrain: 2.1 nTPM (limit 2); pancreas: 3.2 nTPM (limit 2); retina: 2.1 nTPM (limit 2); adrenal gland: 3.4 nTPM (baseline 2); choroid plexus: 32.4 nTPM (baseline 2); hypothalamus: 2.7 nTPM ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 11/50 (22.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 12/176 (6.8%); Testicular Germ Cell Tumor (TCGA): 7/133 (5.3%)

Cancer IHC. liver cancer: 4/11 (36.4%); urothelial cancer: 2/11 (18.2%)

Biology. Receptor tyrosine kinase which binds promiscuously GPI-anchored ephrin-A family ligands residing on adjacent cells, leading to contact-dependent bidirectional signaling into neighboring cells. The signaling pathway downstream of the receptor is referred to as forward signaling while the signaling pathway downstream of the ephrin ligand is referred to as reverse signaling. The GPI-anchored ephrin-A EFNA2, EFNA3, and EFNA5 are able to ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P29322 | HPA portal (live; not the pinned values)

EPHB1 | Unsafe / Protein

ENSG00000154928

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); cerebellum / granular cells - cytoplasm/membrane: Low (Enhanced); cerebellum / processes in granular layer: Low (Enhanced); cerebellum / processes in molecular layer: Medium (Enhanced); cerebellum / white matter cells - ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 50/58 (86.2%); Glioblastoma Multiforme (TCGA): 93/141 (66.0%); Colon Adenocarcinoma (validation): 161/486 (33.1%)

Cancer IHC. renal cancer: 1/11 (9.1%); ovarian cancer: 1/12 (8.3%)

Biology. Receptor tyrosine kinase which binds promiscuously transmembrane ephrin-B family ligands residing on adjacent cells, leading to contact-dependent bidirectional signaling into neighboring cells. The signaling pathway downstream of the receptor is referred to as forward signaling while the signaling pathway downstream of the ephrin ligand is referred to as reverse signaling. Cognate/functional ephrin ligands for this receptor include ... [full text in table]

Curated tissue note. Preferentially expressed in brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P54762 | HPA portal (live; not the pinned values)

EPPIN | Unsafe / High RNA

ENSG00000101448

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 30.9 nTPM; lung/ciliated cells; hpa_single_cell_tissue; vital; 4.7 TPM; blood/Progenitor cell; hpa_immune_monaco; vital

Cancer RNA. Rectum Adenocarcinoma (validation): 55/207 (26.6%); Colon Adenocarcinoma (validation): 103/486 (21.2%); Pancreatic Adenocarcinoma (validation): 9/80 (11.2%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Serine protease inhibitor that plays an essential role in male reproduction and fertility. Modulates the hydrolysis of SEMG1 by KLK3/PSA (a serine protease), provides antimicrobial protection for spermatozoa in the ejaculate coagulum, and binds SEMG1 thereby inhibiting sperm motility

Curated tissue note. In testis, expressed and secreted by Sertoli cells, appearing on the surface of testicular and ejaculate spermatozoa. Expressed in the spermatogonia and the earliest preleptotene spermatocytes. In the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95925 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ERCC6L | Unsafe / Protein

ENSG00000186871

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebral cortex / endothelial cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); cerebral cortex / neuropil: ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 114/133 (85.7%); Colon Adenocarcinoma (validation): 361/486 (74.3%); Breast Invasive Carcinoma (validation): 36/50 (72.0%)

Cancer IHC. prostate cancer: 9/10 (90.0%); breast cancer: 8/9 (88.9%)

Biology. DNA helicase that acts as a tension sensor that associates with catenated DNA which is stretched under tension until it is resolved during anaphase (PubMed:17218258, PubMed:23973328). Functions as ATP-dependent DNA translocase (PubMed:23973328, PubMed:28977671). Can promote Holliday junction branch migration (in vitro) (PubMed:23973328)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q2NKX8 | HPA portal (live; not the pinned values)

ERCC8 | Unsafe / RNA

ENSG00000049167

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3 nTPM (limit 2); amygdala: 3.4 nTPM (limit 2); basal ganglia: 3.3 nTPM (limit 2); bone marrow: 2.6 nTPM (limit 2); cerebral cortex: 2.8 nTPM (limit 2); duodenum: 2.5 nTPM (limit 2); heart muscle: 3.4 nTPM (limit 2); hippocampal formation: 3 nTPM (limit 2); hypothalamus: 3.2 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 76/80 (95.0%); Breast Invasive Carcinoma (validation): 46/50 (92.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Substrate-recognition component of the CSA complex, a DCX (DDB1-CUL4-X-box) E3 ubiquitin-protein ligase complex, involved in transcription-coupled nucleotide excision repair (TC-NER), a process during which RNA polymerase II-blocking lesions are rapidly removed from the transcribed strand of active genes (PubMed:12732143, PubMed:16751180, PubMed:16964240, PubMed:32142649, PubMed:34526721, PubMed:38316879, PubMed:38600235, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13216 | HPA portal (live; not the pinned values)

ERICH2 | Unsafe / RNA

ENSG00000204334

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.2 nTPM (limit 2); amygdala: 4 nTPM (limit 2); basal ganglia: 3.4 nTPM (limit 2); cerebral cortex: 3.9 nTPM (limit 2); choroid plexus: 46.7 nTPM (limit 2); heart muscle: 2.7 nTPM (limit 2); hippocampal formation: 2.9 nTPM (limit 2); hypothalamus: 4.4 nTPM (limit 2); kidney: 3.7 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 51/58 (87.9%); Lung Adenocarcinoma (validation): 87/105 (82.9%); Glioblastoma Multiforme (TCGA): 101/141 (71.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A1L162 | HPA portal (live; not the pinned values)

ERO1B | Unsafe / Protein

ENSG00000086619

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Kidney Renal Clear Cell Carcinoma (validation): 96/100 (96.0%); Lung Squamous Cell Carcinoma (validation): 62/68 (91.2%)

Cancer IHC. testis cancer: 12/12 (100.0%); prostate cancer: 9/10 (90.0%)

Biology. Oxidoreductase involved in disulfide bond formation in the endoplasmic reticulum. Efficiently reoxidizes P4HB/PDI, the enzyme catalyzing protein disulfide formation, in order to allow P4HB to sustain additional rounds of disulfide formation. Other protein disulfide isomerase family members can also be reoxidized, but at lower rates compared to P4HB, including PDIA2 (50% of P4HB reoxidation rate), as well as PDIA3, PDIA4, PDIA6 and ... [full text in table]

Curated tissue note. Highly expressed in the digestive tract, including the duodenum and lower digestive tract. In the stomach, highly expressed in enzyme-producing chief cells (at protein level). In the pancreas, expressed in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86YB8 | HPA portal (live; not the pinned values)

ERVFRD-1 | Unsafe / RNA

ENSG00000244476

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. parathyroid gland: 7.2 nTPM (limit 6); parathyroid gland: 7.2 nTPM (baseline 6)

Cancer RNA. Kidney Chromophobe (TCGA): 7/64 (10.9%); Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%); Bladder Urothelial Carcinoma (TCGA): 3/169 (1.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. This endogenous retroviral envelope protein has retained its original fusogenic properties and participates in trophoblast fusion and the formation of a syncytium during placenta morphogenesis. The interaction with MFSD2A is apparently important for this process (PubMed:18988732) Endogenous envelope proteins may have kept, lost or modified their original function during evolution but this one can still make pseudotypes with MLV, HIV-1 ... [full text in table]

Curated tissue note. Expressed at higher level in placenta. Expressed at lower level in adrenal, bone marrow, brain, breast, colon, kidney, lung, ovary, peripheral blood lymphocytes, prostate, skin, spleen, testis, thymus, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P60508 | HPA portal (live; not the pinned values)

ERVW-1 | Unsafe / RNA

ENSG00000242950

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.4 nTPM (limit 2); retina: 6.7 nTPM (limit 2); choroid plexus: 2.4 nTPM (baseline 2); retina: 6.7 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 80/133 (60.2%); Rectum Adenocarcinoma (validation): 109/207 (52.7%); Colon Adenocarcinoma (validation): 240/486 (49.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. This endogenous retroviral envelope protein has retained its original fusogenic properties and participates in trophoblast fusion and the formation of a syncytium during placenta morphogenesis. May induce fusion through binding of SLC1A4 and SLC1A5 (PubMed:10708449, PubMed:12050356, PubMed:23492904) Endogenous envelope proteins may have kept, lost or modified their original function during evolution. Retroviral envelope proteins ... [full text in table]

Curated tissue note. Expressed at higher level in placental syncytiotrophoblast. Expressed at intermediate level in testis. Seems also to be found at low level in adrenal tissue, bone marrow, breast, colon, kidney, ovary, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UQF0 | HPA portal (live; not the pinned values)

ESCO2 | Unsafe / RNA

ENSG00000171320

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 5.9 nTPM (limit 2); duodenum: 10.7 nTPM (limit 2); small intestine: 7.6 nTPM (limit 2); bone marrow: 5.9 nTPM (baseline 2); duodenum: 10.7 nTPM (baseline 2); small intestine: 7.6 nTPM (baseline 2)

Cancer RNA. Colon Adenocarcinoma (validation): 450/486 (92.6%); Rectum Adenocarcinoma (validation): 176/207 (85.0%); Glioblastoma Multiforme (validation): 46/58 (79.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Acetyltransferase required for the establishment of sister chromatid cohesion (PubMed:15821733, PubMed:15958495). Couples the processes of cohesion and DNA replication to ensure that only sister chromatids become paired together. In contrast to the structural cohesins, the deposition and establishment factors are required only during the S phase. Acetylates the cohesin component SMC3 (PubMed:21111234)

Curated tissue note. Widely expressed in fetal tissues. In adult, it is expressed in thymus, placenta and small intestine

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q56NI9 | HPA portal (live; not the pinned values)

ESPN | Unsafe / Protein

ENSG00000187017

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / Purkinje cells: Medium (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); colon / glandular cells: High (Enhanced); duodenum / glandular cells: High (Enhanced); esophagus / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 205/207 (99.0%); Liver Hepatocellular Carcinoma (TCGA): 355/362 (98.1%); Ovary Serous Cystadenocarcinoma (TCGA): 342/349 (98.0%)

Cancer IHC. liver cancer: 4/10 (40.0%); pancreatic cancer: 2/11 (18.2%)

Biology. Multifunctional actin-bundling protein. Plays a major role in regulating the organization, dimension, dynamics and signaling capacities of the actin filament-rich microvilli in the mechanosensory and chemosensory cells (PubMed:29572253). Required for the assembly and stabilization of the stereociliary parallel actin bundles. Plays a crucial role in the formation and maintenance of inner ear hair cell stereocilia (By similarity). ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt B1AK53 | HPA portal (live; not the pinned values)

ESPNL | Unsafe / RNA

ENSG00000144488

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 4.1 nTPM (limit 2); small intestine: 3.4 nTPM (limit 2); cerebellum: 4.1 nTPM (baseline 2); small intestine: 3.4 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 6/50 (12.0%); Ovary Serous Cystadenocarcinoma (TCGA): 36/349 (10.3%); Thyroid Carcinoma (TCGA): 42/495 (8.5%)

Cancer IHC. breast cancer: 5/11 (45.5%); lung cancer: 3/12 (25.0%)

Biology. Binds to but does not cross-link actin. Required for the formation and maintenance of inner ear hair cell stereocilia and staircase formation. Essential for normal hearing

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZVH7 | HPA portal (live; not the pinned values)

ESR2 | Unsafe / Protein

ENSG00000140009

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / cells in zona glomerulosa: Medium (Enhanced); adrenal gland / glandular cells: Low (Enhanced); adrenal gland / medullary cells: Low (Enhanced); lymph node / non-germinal center cells: Low (Enhanced); rectum / glandular cells: Low (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (validation): 42/207 (20.3%); Colon Adenocarcinoma (validation): 81/486 (16.7%); Glioblastoma Multiforme (validation): 6/58 (10.3%)

Cancer IHC. colorectal cancer: 2/12 (16.7%); cervical cancer: 1/11 (9.1%)

Biology. Nuclear hormone receptor. Binds estrogens with an affinity similar to that of ESR1/ER-alpha, and activates expression of reporter genes containing estrogen response elements (ERE) in an estrogen-dependent manner (PubMed:20074560) Lacks ligand binding ability and has no or only very low ERE binding activity resulting in the loss of ligand-dependent transactivation ability

Curated tissue note. Expressed in testis and ovary, and at a lower level in heart, brain, placenta, liver, skeletal muscle, spleen, thymus, prostate, colon, bone marrow, mammary gland and uterus. Also found in uterine bone, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92731 | HPA portal (live; not the pinned values)

ESYT2 | Unsafe / Protein

ENSG00000117868

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebral cortex / endothelial cells: Medium (Approved); cerebral cortex / glial cells: Low (Approved); cerebral cortex / neuronal ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%)

Cancer IHC. colorectal cancer: 8/12 (66.7%); prostate cancer: 4/10 (40.0%)

Biology. Tethers the endoplasmic reticulum to the cell membrane and promotes the formation of appositions between the endoplasmic reticulum and the cell membrane. Binds glycerophospholipids in a barrel-like domain and may play a role in cellular lipid transport. Plays a role in FGF signaling via its role in the rapid internalization of FGFR1 that has been activated by FGF1 binding; this occurs most likely via the AP-2 complex. Promotes the ... [full text in table]

Curated tissue note. Widely expressed with high level in cerebellum

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0FGR8 | HPA portal (live; not the pinned values)

ETFBKMT | Unsafe / Protein

ENSG00000139160

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / Purkinje cells: Medium (Uncertain); cerebral cortex / glial cells: Low (Uncertain); cerebral cortex / neuronal cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 54/58 (93.1%); Testicular Germ Cell Tumor (TCGA): 71/133 (53.4%); Prostate Adenocarcinoma (TCGA): 169/480 (35.2%)

Cancer IHC. renal cancer: 10/12 (83.3%); prostate cancer: 8/11 (72.7%)

Biology. Protein-lysine methyltransferase that selectively trimethylates the flavoprotein ETFB in mitochondria (PubMed:25023281, PubMed:25416781). Thereby, may negatively regulate the function of ETFB in electron transfer from Acyl-CoA dehydrogenases to the main respiratory chain (PubMed:25416781)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IXQ9 | HPA portal (live; not the pinned values)

ETFRF1 | Unsafe / Protein

ENSG00000205707

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / endothelial cells: Medium (Approved); esophagus / squamous epithelial cells: Low (Approved); heart muscle / cardiomyocytes: Medium (Approved); lung / alveolar cells: Low (Approved); oral mucosa / squamous epithelial cells: Medium (Approved); skeletal muscle / myocytes: Low (Approved); skin ... [full text in table]

Cancer RNA. Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Acts as a regulator of the electron transfer flavoprotein by promoting the removal of flavin from the ETF holoenzyme (composed of ETFA and ETFB)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6IPR1 | HPA portal (live; not the pinned values)

ETNK1 | Unsafe / Protein

ENSG00000139163

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Medium (Approved); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. cervical cancer: 3/12 (25.0%); ovarian cancer: 3/12 (25.0%)

Biology. Highly specific for ethanolamine phosphorylation. May be a rate-controlling step in phosphatidylethanolamine biosynthesis

Curated tissue note. Expressed in kidney, liver, placenta, heart, leukocyte, ovary and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HBU6 | HPA portal (live; not the pinned values)

ETV2 | Unsafe / RNA

ENSG00000105672

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. pancreas: 2.3 nTPM (limit 2); pancreas: 2.3 nTPM (baseline 2)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 51/99 (51.5%); Ovary Serous Cystadenocarcinoma (TCGA): 117/349 (33.5%); Bladder Urothelial Carcinoma (TCGA): 45/169 (26.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Binds to DNA sequences containing the consensus pentanucleotide 5'-CGGA[AT]-3'

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00321 | HPA portal (live; not the pinned values)

EXO1 | Unsafe / RNA

ENSG00000174371

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 17.1 nTPM (limit 2); duodenum: 2.8 nTPM (limit 2); small intestine: 2.4 nTPM (limit 2); thymus: 10.8 nTPM (limit 10); bone marrow: 17.1 nTPM (baseline 2); duodenum: 2.8 nTPM (baseline 2); small intestine: 2.4 nTPM (baseline 2); thymus: 10.8 nTPM (baseline 10)

Cancer RNA. Colon Adenocarcinoma (validation): 476/486 (97.9%); Testicular Germ Cell Tumor (TCGA): 128/133 (96.2%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 269/283 (95.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. 5'->3' double-stranded DNA exonuclease which may also possess a cryptic 3'->5' double-stranded DNA exonuclease activity. Functions in DNA mismatch repair (MMR) to excise mismatch-containing DNA tracts directed by strand breaks located either 5' or 3' to the mismatch. Also exhibits endonuclease activity against 5'-overhanging flap structures similar to those generated by displacement synthesis when DNA polymerase encounters the 5'-end ... [full text in table]

Curated tissue note. Highly expressed in bone marrow, testis and thymus. Expressed at lower levels in colon, lymph nodes, ovary, placenta, prostate, small intestine, spleen and stomach

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UQ84 | HPA portal (live; not the pinned values)

F11R | Unsafe / Protein

ENSG00000158769

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebral cortex / ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. lung cancer: 12/12 (100.0%); urothelial cancer: 10/10 (100.0%)

Biology. Seems to play a role in epithelial tight junction formation. Appears early in primordial forms of cell junctions and recruits PARD3 (PubMed:11489913). The association of the PARD6-PARD3 complex may prevent the interaction of PARD3 with JAM1, thereby preventing tight junction assembly (By similarity). Plays a role in regulating monocyte transmigration involved in integrity of epithelial barrier (By similarity). Ligand for integrin ... [full text in table]

Curated tissue note. Expressed in endothelium, epithelium and leukocytes (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y624 | HPA portal (live; not the pinned values)

F2RL2 | Unsafe / RNA

ENSG00000164220

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. midbrain: 2.6 nTPM (limit 2); retina: 4.8 nTPM (limit 2); small intestine: 2.2 nTPM (limit 2); smooth muscle: 3.6 nTPM (limit 2); midbrain: 2.6 nTPM (baseline 2); retina: 4.8 nTPM (baseline 2); small intestine: 2.2 nTPM (baseline 2); smooth muscle: 3.6 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 193/207 (93.2%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Colon Adenocarcinoma (validation): 416/486 (85.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Receptor for activated thrombin coupled to G proteins that stimulate phosphoinositide hydrolysis

Curated tissue note. Highest expression in the megakaryocytes of the bone marrow, lower in mature megakaryocytes, in platelets and in a variety of other tissues such as heart and gut

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00254 | HPA portal (live; not the pinned values)

FABP12 | Unsafe / Protein

ENSG00000197416

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); hippocampus / neuronal cells: Low (Enhanced); nasopharynx / respiratory epithelial cells: Low (Enhanced); skeletal muscle / myocytes: Medium ... [full text in table]

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 15/283 (5.3%); Head and Neck Squamous Cell Carcinoma (TCGA): 6/492 (1.2%); Lung Squamous Cell Carcinoma (TCGA): 4/489 (0.8%)

Cancer IHC. lung cancer: 3/12 (25.0%); cervical cancer: 1/9 (11.1%)

Biology. May play a role in lipid transport

Curated tissue note. Expressed in a number of retinoblastoma cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NFH5 | HPA portal (live; not the pinned values)

FAF1 | Unsafe / Protein

ENSG00000185104

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebellum / cells in molecular layer: Medium (Approved); cerebellum / Purkinje cells: ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. cervical cancer: 9/9 (100.0%); colorectal cancer: 9/9 (100.0%)

Biology. Ubiquitin-binding protein (PubMed:19722279). Required for the progression of DNA replication forks by targeting DNA replication licensing factor CDT1 for degradation (PubMed:26842564). Potentiates but cannot initiate FAS-induced apoptosis (By similarity)

Curated tissue note. Most abundant in testis, slightly less abundant in skeletal muscle and heart, followed by prostate, thymus, ovary, small intestine, and colon. Not detected in the peripheral blood leukocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UNN5 | HPA portal (live; not the pinned values)

FAHD1 | Unsafe / Protein

ENSG00000180185

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / molecular layer cells - cytoplasm/membrane: High (Enhanced); cerebellum / processes ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); breast cancer: 11/12 (91.7%)

Biology. Tautomerase that converts enol-oxaloacetate, a strong inhibitor of succinate dehydrogenase, to the physiological keto form of oxaloacetate (PubMed:38287013). It is thereby required to maximize aerobic respiration efficiency by preventing succinate dehydrogenase inhibition (PubMed:38287013). Also acts as a weak oxaloacetate decarboxylase (ODx), catalyzing the decarboxylation of oxaloacetate (OAA) to pyruvate and CO(2), and as such is ... [full text in table]

Curated tissue note. Ubiquitous (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6P587 | HPA portal (live; not the pinned values)

FAM118B | Unsafe / Protein

ENSG00000197798

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: High (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / glial cells: High (Approved); cerebral ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. glioma: 9/11 (81.8%); stomach cancer: 7/11 (63.6%)

Biology. May play a role in Cajal bodies formation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BPY3 | HPA portal (live; not the pinned values)

FAM133A | Unsafe / RNA

ENSG00000179083

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.7 nTPM (limit 2); basal ganglia: 4.7 nTPM (limit 2); cerebellum: 2.2 nTPM (limit 2); cerebral cortex: 3.1 nTPM (limit 2); hippocampal formation: 2.3 nTPM (limit 2); hypothalamus: 4.6 nTPM (limit 2); midbrain: 2.3 nTPM (limit 2); pituitary gland: 2.1 nTPM (limit 2); spinal cord: 2.4 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 21/58 (36.2%); Testicular Germ Cell Tumor (TCGA): 27/133 (20.3%); Liver Hepatocellular Carcinoma (validation): 36/231 (15.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N9E0 | HPA portal (live; not the pinned values)

FAM161A | Unsafe / Protein

ENSG00000170264

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): Medium (Approved); bronchus / ciliated cells (ciliary rootlets): High (Approved); bronchus / ciliated cells (tip of cilia): High (Approved); cerebral cortex / endothelial cells: Low (Approved); kidney / cells in glomeruli: Low (Approved); lung / alveolar ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 42/50 (84.0%); Ovary Serous Cystadenocarcinoma (validation): 23/81 (28.4%)

Cancer IHC. colorectal cancer: 5/12 (41.7%); pancreatic cancer: 2/12 (16.7%)

Biology. Involved in ciliogenesis

Curated tissue note. Isoform 1 and isoform 3 are widely expressed with highest levels in retina and testis, with isoform 1 being the most abundant in all tissues tested

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3B820 | HPA portal (live; not the pinned values)

FAM163A | Unsafe / RNA

ENSG00000143340

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 5.8 nTPM (limit 2); cerebral cortex: 2.4 nTPM (limit 2); pituitary gland: 28.8 nTPM (limit 2); thymus: 15.6 nTPM (limit 10); adrenal gland: 5.8 nTPM (baseline 2); cerebral cortex: 2.4 nTPM (baseline 2); pituitary gland: 28.8 nTPM (baseline 2); thymus: 15.6 nTPM (baseline 10)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 8/99 (8.1%); Uterine Corpus Endometrial Carcinoma (TCGA): 9/176 (5.1%); Pancreatic Adenocarcinoma (TCGA): 7/176 (4.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Highly expressed in neuroblastoma compared to other tissues, suggesting that it may be used as a marker for metastasis in bone marrow

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96GL9 | HPA portal (live; not the pinned values)

FAM168B | Unsafe / Protein

ENSG00000152102

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Enhanced); cerebellum / cells in granular layer: Low (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); heart muscle / cardiomyocytes: High (Enhanced); hippocampus / neuronal cells: Low (Enhanced); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%)

Cancer IHC. breast cancer: 0/9 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Inhibitor of neuronal axonal outgrowth. Acts as a negative regulator of CDC42 and STAT3 and a positive regulator of STMN2. Positive regulator of CDC27

Curated tissue note. Expressed in the brain, within neuronal axonal fibers and associated with myelin sheets (at protein level). Expression tends to be lower in the brain of Alzheimer disease patients compared to healthy ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A1KXE4 | HPA portal (live; not the pinned values)

FAM181A | Unsafe / RNA

ENSG00000140067

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 13.5 nTPM (limit 2); basal ganglia: 9.9 nTPM (limit 2); hippocampal formation: 8.9 nTPM (limit 2); hypothalamus: 9.2 nTPM (limit 2); midbrain: 8.1 nTPM (limit 2); pituitary gland: 2.2 nTPM (limit 2); spinal cord: 4.4 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (TCGA): 116/141 (82.3%); Glioblastoma Multiforme (validation): 39/58 (67.2%); Ovary Serous Cystadenocarcinoma (validation): 46/81 (56.8%)

Cancer IHC. ovarian cancer: 1/11 (9.1%); breast cancer: 0/9 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N9Y4 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

FAM209A | Unsafe / RNA

ENSG00000124103

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 16.5 nTPM (limit 2); bone marrow: 16.5 nTPM (baseline 2)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Bladder Urothelial Carcinoma (TCGA): 2/169 (1.2%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May play a role in sperm acrosome biogenesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JX71 | HPA portal (live; not the pinned values)

FAM209B | Unsafe / High RNA

ENSG00000213714

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 11.6 nTPM; bone marrow/bulk; hpa_rna_consensus; vital; 30.91 TPM; blood/NK; dice_nk; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 7/283 (2.5%); Kidney Chromophobe (TCGA): 1/64 (1.6%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May play a role in sperm acrosome biogenesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JX69 | HPA portal (live; not the pinned values)

FAM221B | Unsafe / Protein

ENSG00000204930

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / cells in tubules: Low (Enhanced); lung / alveolar cells: Low (Enhanced); parathyroid gland / glandular cells: Low (Enhanced); spleen / cells in red pulp: Low (Enhanced); stomach 2 / glandular cells: Low (Enhanced)

Cancer RNA. Kidney Chromophobe (TCGA): 17/64 (26.6%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 20/283 (7.1%); Skin Cuteneous Melanoma (TCGA): 4/99 (4.0%)

Cancer IHC. prostate cancer: 8/12 (66.7%); renal cancer: 2/11 (18.2%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6H8Z2 | HPA portal (live; not the pinned values)

FAM229A | Unsafe / RNA

ENSG00000225828

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 6.5 nTPM (limit 2); amygdala: 7.4 nTPM (limit 2); basal ganglia: 6.2 nTPM (limit 2); bone marrow: 2.1 nTPM (limit 2); cerebellum: 8 nTPM (limit 2); cerebral cortex: 5.6 nTPM (limit 2); heart muscle: 3.2 nTPM (limit 2); hippocampal formation: 5.7 nTPM (limit 2); hypothalamus: 5.5 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 58/141 (41.1%); Glioblastoma Multiforme (validation): 20/58 (34.5%); Ovary Serous Cystadenocarcinoma (TCGA): 118/349 (33.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt H3BQW9 | HPA portal (live; not the pinned values)

FAM229B | Unsafe / RNA

ENSG00000203778

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 17.6 nTPM (limit 10); adrenal gland: 21 nTPM (limit 2); amygdala: 74.3 nTPM (limit 2); basal ganglia: 72 nTPM (limit 2); bone marrow: 3.7 nTPM (limit 2); cerebellum: 23.4 nTPM (limit 2); cerebral cortex: 58.9 nTPM (limit 2); choroid plexus: 101.6 nTPM (limit 2); colon: 20.4 nTPM ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 495/495 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4G0N7 | HPA portal (live; not the pinned values)

FAM25C | Unsafe / RNA

ENSG00000276430

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. esophagus: 29.3 nTPM (limit 6); salivary gland: 14.9 nTPM (limit 6); skin: 42.2 nTPM (limit 10); esophagus: 29.3 nTPM (baseline 6); salivary gland: 14.9 nTPM (baseline 6); skin: 42.2 nTPM (baseline 10)

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 251/492 (51.0%); Skin Cuteneous Melanoma (TCGA): 17/99 (17.2%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 40/283 (14.1%)

Cancer IHC. liver cancer: 12/12 (100.0%); testis cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt B3EWG3 | UniProt B3EWG5 | UniProt B3EWG6 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

FAM25G | Unsafe / RNA

ENSG00000189090

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. esophagus: 18.1 nTPM (limit 6); salivary gland: 8.6 nTPM (limit 6); skin: 41.9 nTPM (limit 10); esophagus: 18.1 nTPM (baseline 6); salivary gland: 8.6 nTPM (baseline 6); skin: 41.9 nTPM (baseline 10)

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 50/492 (10.2%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Bladder Urothelial Carcinoma (TCGA): 2/169 (1.2%)

Cancer IHC. liver cancer: 12/12 (100.0%); testis cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt B3EWG3 | UniProt B3EWG5 | UniProt B3EWG6 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

FAM72A | Unsafe / RNA

ENSG00000196550

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.1 nTPM (limit 2); amygdala: 2.1 nTPM (limit 2); bone marrow: 8.5 nTPM (limit 2); cerebellum: 5.5 nTPM (limit 2); duodenum: 2.9 nTPM (limit 2); hippocampal formation: 2.4 nTPM (limit 2); lung: 2.6 nTPM (limit 2); lymph node: 10.7 nTPM (limit 6); midbrain: 2.6 nTPM (limit 2); small ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 440/486 (90.5%); Rectum Adenocarcinoma (validation): 184/207 (88.9%); Lung Squamous Cell Carcinoma (validation): 54/68 (79.4%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May play a role in the regulation of cellular reactive oxygen species metabolism. May participate in cell growth regulation

Curated tissue note. May be up-regulated in malignant colon cancers, compared to normal colon and colon adenomas. Expression is also elevated in other common cancer types, including breast, lung, uterus, and ovary

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5TYM5 | HPA portal (live; not the pinned values)

FAM76B | Unsafe / Protein

ENSG00000077458

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. lymph node / germinal center cells: Low (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 206/207 (99.5%); Colon Adenocarcinoma (validation): 479/486 (98.6%)

Cancer IHC. glioma: 4/12 (33.3%); breast cancer: 0/12 (0.0%)

Biology. Negatively regulates the NF-kappa-B-mediated inflammatory pathway by preventing the translocation of HNRNPA2B1 from the nucleus to the cytoplasm (PubMed:37643469). Inhibits the PI3K/Akt/NF-kappa-B pathway-mediated polarization of M1 macrophages by binding to and stabilizing PIK3CD mRNA, resulting in increased levels of PIK3CD protein and increased levels of phosphorylated downstream target AKT which leads to decreased NF-kappa-B ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5HYJ3 | HPA portal (live; not the pinned values)

FAM78B | Unsafe / High RNA

ENSG00000188859

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 171.5 nTPM; brain/inhibitory neurons; hpa_single_cell_tissue; vital; 3.496 TPM; blood/TFH; dice_tfh; vital

Cancer RNA. Glioblastoma Multiforme (validation): 31/58 (53.4%); Breast Invasive Carcinoma (validation): 22/50 (44.0%); Skin Cuteneous Melanoma (TCGA): 16/99 (16.2%)

Cancer IHC. melanoma: 8/8 (100.0%); colorectal cancer: 9/10 (90.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VT40 | HPA portal (live; not the pinned values)

FAM81B | Unsafe / RNA

ENSG00000153347

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.2 nTPM (limit 2); basal ganglia: 4.2 nTPM (limit 2); choroid plexus: 60.6 nTPM (limit 2); heart muscle: 7.3 nTPM (limit 4); hippocampal formation: 5.1 nTPM (limit 2); hypothalamus: 6.7 nTPM (limit 2); lung: 4.2 nTPM (limit 4); pituitary gland: 3.6 nTPM (limit 2)

Cancer RNA. Kidney Renal Papillary Cell Carcinoma (TCGA): 97/282 (34.4%); Uterine Corpus Endometrial Carcinoma (TCGA): 45/176 (25.6%); Lung Adenocarcinoma (TCGA): 124/497 (24.9%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96LP2 | HPA portal (live; not the pinned values)

FAM83F | Unsafe / Protein

ENSG00000133477

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / endothelial cells: Medium (Enhanced); colon / glandular cells: Medium (Enhanced); colon / peripheral nerve/ganglion: Low (Enhanced); duodenum / glandular cells: High (Enhanced); heart muscle / cardiomyocytes: Low (Enhanced); kidney / cells in glomeruli: Low (Enhanced); kidney / cells in ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 205/207 (99.0%); Colon Adenocarcinoma (validation): 481/486 (99.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 478/492 (97.2%)

Cancer IHC. colorectal cancer: 6/11 (54.5%); ovarian cancer: 3/12 (25.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEG4 | HPA portal (live; not the pinned values)

FAM90A1 | Unsafe / RNA

ENSG00000171847

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 2.2 nTPM (limit 2); cerebral cortex: 6.8 nTPM (limit 2); retina: 4.5 nTPM (limit 2); cerebellum: 2.2 nTPM (baseline 2); cerebral cortex: 6.8 nTPM (baseline 2); retina: 4.5 nTPM (baseline 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 174/349 (49.9%); Glioblastoma Multiforme (TCGA): 70/141 (49.6%); Testicular Germ Cell Tumor (TCGA): 47/133 (35.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86YD7 | HPA portal (live; not the pinned values)

FAM9B | Unsafe / RNA

ENSG00000177138

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. liver: 3 nTPM (limit 2); liver: 3 nTPM (baseline 2)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 22/231 (9.5%); Testicular Germ Cell Tumor (TCGA): 12/133 (9.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 7/176 (4.0%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); ovarian cancer: 11/12 (91.7%)

Biology. May play a role in meiosis

Curated tissue note. Expressed in testis and ovary (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZU0 | HPA portal (live; not the pinned values)

FAM9C | Unsafe / RNA

ENSG00000187268

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. spleen: 37.2 nTPM (limit 10); spleen: 37.2 nTPM (baseline 10)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 67/133 (50.4%); Bladder Urothelial Carcinoma (TCGA): 8/169 (4.7%); Pancreatic Adenocarcinoma (validation): 2/80 (2.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Expressed exclusively in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZT9 | HPA portal (live; not the pinned values)

FANCA | Unsafe / RNA

ENSG00000187741

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.1 nTPM (limit 2); bone marrow: 11.3 nTPM (limit 2); cerebellum: 5.2 nTPM (limit 2); cerebral cortex: 3.3 nTPM (limit 2); duodenum: 3.4 nTPM (limit 2); esophagus: 6.4 nTPM (limit 6); kidney: 2.8 nTPM (limit 2); liver: 5.9 nTPM (limit 2); lung: 3.2 nTPM (limit 2); lymph node: 7.7 ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 485/486 (99.8%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 279/283 (98.6%); Rectum Adenocarcinoma (validation): 203/207 (98.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. DNA repair protein that may operate in a postreplication repair or a cell cycle checkpoint function. May be involved in interstrand DNA cross-link repair and in the maintenance of normal chromosome stability

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O15360 | HPA portal (live; not the pinned values)

FANCM | Unsafe / RNA

ENSG00000187790

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 3.3 nTPM (limit 2); bone marrow: 3.3 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Colon Adenocarcinoma (validation): 219/486 (45.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. DNA-dependent ATPase component of the Fanconi anemia (FA) core complex (PubMed:16116422). Required for the normal activation of the FA pathway, leading to monoubiquitination of the FANCI-FANCD2 complex in response to DNA damage, cellular resistance to DNA cross-linking drugs, and prevention of chromosomal breakage (PubMed:16116422, PubMed:19423727, PubMed:20347428, PubMed:20347429, PubMed:29231814). In complex with CENPS and CENPX, ... [full text in table]

Curated tissue note. Expressed in germ cells of fetal and adult ovaries. In fetal ovaries, it is present in oogonia but expression is stronger in pachytene stage oocytes. Expressed in oocytes arrested at the diplotene stage of ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYD8 | HPA portal (live; not the pinned values)

FANK1 | Unsafe / RNA

ENSG00000203780

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.5 nTPM (limit 2); amygdala: 5.2 nTPM (limit 2); basal ganglia: 7.4 nTPM (limit 2); cerebral cortex: 2.9 nTPM (limit 2); choroid plexus: 66.4 nTPM (limit 2); duodenum: 2.3 nTPM (limit 2); hippocampal formation: 6.9 nTPM (limit 2); hypothalamus: 9.5 nTPM (limit 2); kidney: 5.7 nTPM ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 474/495 (95.8%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 207/282 (73.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Through the activation of JUN and AP-1-mediated transcription, may regulate apoptosis

Curated tissue note. Mostly restricted to testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TC84 | HPA portal (live; not the pinned values)

FATE1 | Unsafe / High RNA

ENSG00000147378

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 41.2 nTPM; small intestine/enteroendocrine cells; hpa_single_cell_tissue; vital; 0.0064 TPM; blood/naive CD8 T-cell; hpa_immune_cell_samples; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 27/133 (20.3%); Kidney Renal Clear Cell Carcinoma (TCGA): 102/521 (19.6%); Kidney Renal Clear Cell Carcinoma (validation): 14/100 (14.0%)

Cancer IHC. ovarian cancer: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. Involved in the regulation of endoplasmic reticulum (ER)-mitochondria coupling. Negatively regulates the ER-mitochondria distance and Ca(2+) transfer from ER to mitochondria possibly implicating it in the regulation of apoptosis (PubMed:27402544). May collaborate with RNF183 to restrain BIK protein levels thus regulating apoptotic signaling (PubMed:26567849)

Curated tissue note. Testis-specific in fetus (aged from 6 to 11 weeks). In adult, expressed predominantly in testis, with some expression in lung, heart, kidney, adrenal gland and whole brain (PubMed:11694338). Highly expressed ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q969F0 | HPA portal (live; not the pinned values)

FBXL13 | Unsafe / Protein

ENSG00000161040

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / Purkinje cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 50/58 (86.2%); Pancreatic Adenocarcinoma (validation): 17/80 (21.2%); Breast Invasive Carcinoma (validation): 3/50 (6.0%)

Cancer IHC. liver cancer: 3/11 (27.3%); glioma: 1/8 (12.5%)

Biology. Substrate-recognition component of the SCF (SKP1-CUL1-F-box protein)-type E3 ubiquitin ligase complex. Component of the nexin-dynein regulatory complex (N-DRC), a key regulator of ciliary/flagellar motility which maintains the alignment and integrity of the distal axoneme and regulates microtubule sliding in motile axonemes. Specifically targets CEP192 isoform 3 for ubiquitin-mediated proteolysis and thereby acts as a regulator of ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEE6 | HPA portal (live; not the pinned values)

FBXL17 | Unsafe / Protein

ENSG00000145743

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 105/105 (100.0%); Kidney Chromophobe (TCGA): 64/64 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); skin cancer: 12/12 (100.0%)

Biology. Substrate-recognition component of the SCF(FBXL17) E3 ubiquitin ligase complex, a key component of a quality control pathway required to ensure functional dimerization of BTB domain-containing proteins (dimerization quality control, DQC) (PubMed:30190310). FBXL17 specifically recognizes and binds a conserved degron of non-consecutive residues present at the interface of BTB dimers of aberrant composition: aberrant BTB dimer are then ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UF56 | HPA portal (live; not the pinned values)

FBXO10 | Unsafe / Protein

ENSG00000147912

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / glandular cells: Medium (Uncertain); duodenum / glandular cells: Medium (Uncertain); lung / macrophages: Low (Uncertain); rectum / glandular cells: Medium (Uncertain); small intestine / glandular cells: Medium (Uncertain)

Cancer RNA. Glioblastoma Multiforme (validation): 49/58 (84.5%); Breast Invasive Carcinoma (validation): 40/50 (80.0%); Testicular Germ Cell Tumor (TCGA): 105/133 (78.9%)

Cancer IHC. renal cancer: 4/12 (33.3%); liver cancer: 2/10 (20.0%)

Biology. Substrate-recognition component of the SCF (SKP1-CUL1-F-box protein)-type E3 ubiquitin ligase complex. Mediates the ubiquitination and degradation of BCL2, an antiapoptotic protein, thereby playing a role in apoptosis by controlling the stability of BCL2. Targets also the receptor for advanced glycation end products RAGE for ubiquitination and subsequent lysosomal degradation (PubMed:28515150). Directly controls HGAL/GCSAM ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UK96 | HPA portal (live; not the pinned values)

FBXO15 | Unsafe / RNA

ENSG00000141665

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.9 nTPM (limit 2); retina: 3.3 nTPM (limit 2); choroid plexus: 2.9 nTPM (baseline 2); retina: 3.3 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 35/50 (70.0%); Pancreatic Adenocarcinoma (validation): 6/80 (7.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 21/282 (7.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Substrate-recognition component of the SCF (SKP1-CUL1-F-box protein)-type E3 ubiquitin ligase complex

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCQ5 | HPA portal (live; not the pinned values)

FBXO38 | Unsafe / Protein

ENSG00000145868

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); cerebral cortex / glial cells: Low (Approved); cerebral cortex / neuronal cells: Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); pancreatic cancer: 8/10 (80.0%)

Biology. Substrate recognition component of a SCF (SKP1-CUL1-F-box protein) E3 ubiquitin-protein ligase complex which mediates the ubiquitination and subsequent proteasomal degradation of PDCD1/PD-1, thereby regulating T-cells-mediated immunity (PubMed:30487606). Required for anti-tumor activity of T-cells by promoting the degradation of PDCD1/PD-1; the PDCD1-mediated inhibitory pathway being exploited by tumors to attenuate anti-tumor immunity ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PIJ6 | HPA portal (live; not the pinned values)

FBXO5 | Unsafe / Protein

ENSG00000112029

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 482/486 (99.2%)

Cancer IHC. pancreatic cancer: 7/9 (77.8%); ovarian cancer: 7/11 (63.6%)

Biology. Regulator of APC activity during mitotic and meiotic cell cycle (PubMed:16921029, PubMed:17234884, PubMed:17485488, PubMed:17875940, PubMed:23708001, PubMed:23708605). During mitotic cell cycle plays a role as both substrate and inhibitor of APC-FZR1 complex (PubMed:16921029, PubMed:17234884, PubMed:17485488, PubMed:17875940, PubMed:23708001, PubMed:23708605, PubMed:29875408). During G1 phase, plays a role as substrate of APC-FZR1 ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UKT4 | HPA portal (live; not the pinned values)

FER1L6 | Unsafe / Protein

ENSG00000214814

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); colon / goblet cells: Low (Enhanced); duodenum / goblet cells: Medium (Enhanced); rectum / goblet cells: Medium (Enhanced); skeletal muscle / myocytes: Low (Enhanced); small intestine / goblet ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 51/80 (63.7%); Colon Adenocarcinoma (validation): 219/486 (45.1%); Rectum Adenocarcinoma (validation): 92/207 (44.4%)

Cancer IHC. stomach cancer: 8/12 (66.7%); pancreatic cancer: 7/12 (58.3%)

Biology. Calcium/phospholipid-binding protein which is essential for proper development of skeletal and cardiac muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q2WGJ9 | HPA portal (live; not the pinned values)

FEZF1 | Unsafe / Protein

ENSG00000128610

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Approved); cerebral cortex / neuropil: Medium (Approved); hippocampus / neuronal cells: Low (Approved); hypothalamus / neuronal cells: Low (Approved); hypothalamus / neuronal projections: Medium (Approved)

Cancer RNA. Colon Adenocarcinoma (validation): 178/486 (36.6%); Lung Squamous Cell Carcinoma (TCGA): 153/489 (31.3%); Lung Adenocarcinoma (TCGA): 133/497 (26.8%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Transcription repressor. Involved in the axonal projection and proper termination of olfactory sensory neurons (OSN). Plays a role in rostro-caudal patterning of the diencephalon and in prethalamic formation. Expression is required in OSN to cell-autonomously regulate OSN axon projections. Regulates non-cell-autonomously the layer formation of the olfactory bulb development and the interneurons. May be required for correct rostral ... [full text in table]

Curated tissue note. Expressed in brain. Little or no expression in other tissues. Overexpressed specifically in gastric cancers. A 2- to 20-fold increase is found in over 50% of gastric cancer tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0PJY2 | HPA portal (live; not the pinned values)

FHAD1 | Unsafe / Protein

ENSG00000142621

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (ciliary rootlets): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cell body): Medium (Enhanced); nasopharynx / ciliated cells (cilia axoneme): Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 34/50 (68.0%); Pancreatic Adenocarcinoma (validation): 37/80 (46.2%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 55/283 (19.4%)

Cancer IHC. ovarian cancer: 1/11 (9.1%); breast cancer: 0/11 (0.0%)

Biology. Regulator of sperm motility and spermatocyte meiosis

Curated tissue note. Testis (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt B1AJZ9 | HPA portal (live; not the pinned values)

FIGNL1 | Unsafe / Protein

ENSG00000132436

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. liver cancer: 8/11 (72.7%); breast cancer: 6/11 (54.5%)

Biology. ATPase that maintains genome stability by acting as a key regulator of DNA double-strand break (DSB) repair via homologous recombination (HR) (PubMed:23754376, PubMed:37256941, PubMed:37347663, PubMed:37515771, PubMed:37556550, PubMed:38286805, PubMed:38597669, PubMed:39636933, PubMed:41166468). Regulates HR by mediating disassembly of RAD51 filaments from DNA and chromatin, thereby preventing DNA damage-independent RAD51 loading and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PIW4 | HPA portal (live; not the pinned values)

FIP1L1 | Unsafe / Protein

ENSG00000145216

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Supported); adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: Medium (Supported); bronchus / ciliated cells (cell body): Medium (Supported); caudate / glial cells: Low (Supported); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Ovary Serous Cystadenocarcinoma (validation): 81/81 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Component of the cleavage and polyadenylation specificity factor (CPSF) complex that plays a key role in pre-mRNA 3'-end formation, recognizing the AAUAAA signal sequence and interacting with poly(A) polymerase and other factors to bring about cleavage and poly(A) addition. FIP1L1 contributes to poly(A) site recognition and stimulates poly(A) addition. Binds to U-rich RNA sequence elements surrounding the poly(A) site. May act to ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6UN15 | HPA portal (live; not the pinned values)

FIRRM | Unsafe / Protein

ENSG00000000460

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Rectum Adenocarcinoma (validation): 186/207 (89.9%)

Cancer IHC. cervical cancer: 10/10 (100.0%); skin cancer: 11/12 (91.7%)

Biology. Forms a complex with FIGL1, which regulates DNA double-strand break (DSB) repair via homologous recombination (HR) (PubMed:29608566, PubMed:37256941, PubMed:37347663, PubMed:37515771, PubMed:37556550, PubMed:38286805). The FIGNL1-FIRRM complex is essential for resolution of homologous recombination intermediates in response to DNA interstrand cross-links (ICLs) (PubMed:37256941, PubMed:37347663, PubMed:37515771, PubMed:37556550, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NSG2 | HPA portal (live; not the pinned values)

FLACC1 | Unsafe / RNA

ENSG00000155749

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 5 nTPM (limit 2); choroid plexus: 8.7 nTPM (limit 2); heart muscle: 4.1 nTPM (limit 2); lung: 2.4 nTPM (limit 2); retina: 3.2 nTPM (limit 2); bone marrow: 5 nTPM (baseline 2); choroid plexus: 8.7 nTPM (baseline 2); heart muscle: 4.1 nTPM (baseline 2); lung: 2.4 nTPM (baseline 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 50/58 (86.2%); Rectum Adenocarcinoma (validation): 13/207 (6.3%); Uterine Corpus Endometrial Carcinoma (TCGA): 10/176 (5.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96Q35 | HPA portal (live; not the pinned values)

FMC1 | Unsafe / Protein

ENSG00000164898

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / basal cells: Low (Approved); bronchus / ciliated cells (cell body): Medium (Approved); caudate / glial cells: Medium (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / glial cells: Low (Approved); colon / ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. prostate cancer: 9/10 (90.0%); colorectal cancer: 10/12 (83.3%)

Biology. Plays a role in the assembly/stability of the mitochondrial membrane ATP synthase (F(1)F(0) ATP synthase or Complex V) (PubMed:28719601)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96HJ9 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

FMN1 | Unsafe / Protein

ENSG00000248905

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Supported); bronchus / ciliated cells (cell body): Medium (Supported); colon / endothelial cells: Medium (Supported); colon / glandular cells: Medium (Supported); duodenum / glandular cells: Medium (Supported); esophagus / squamous epithelial cells: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. colorectal cancer: 10/12 (83.3%); pancreatic cancer: 10/12 (83.3%)

Biology. Plays a role in the formation of adherens junction and the polymerization of linear actin cables

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q68DA7 | HPA portal (live; not the pinned values)

FNDC11 | Unsafe / High RNA

ENSG00000125531

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 37.4 nTPM; bronchus/ciliated cells; hpa_single_cell_tissue; vital; 8.129 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 52/349 (14.9%); Bladder Urothelial Carcinoma (TCGA): 22/169 (13.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 16/176 (9.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BVV2 | HPA portal (live; not the pinned values)

FOXB1 | Unsafe / RNA

ENSG00000171956

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. hypothalamus: 3.3 nTPM (limit 2); midbrain: 4.1 nTPM (limit 2); hypothalamus: 3.3 nTPM (baseline 2); midbrain: 4.1 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 18/176 (10.2%); Ovary Serous Cystadenocarcinoma (TCGA): 21/349 (6.0%); Ovary Serous Cystadenocarcinoma (validation): 3/81 (3.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcription factor expressed by neural progenitor cells in specific regions of the embryonic neuroepithelium. Essential for the mammillary nuclei maintenance. Negatively regulates the proliferation of oligodendrocyte progenitors and promotes oligodendrocyte maturation. Also expressed in mammary glands, plays a role in lactation, controls development of mammary glands and the inferior colliculi of the midbrain in the central nervous ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q99853 | HPA portal (live; not the pinned values)

FOXD1 | Unsafe / RNA

ENSG00000251493

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. hypothalamus: 2.7 nTPM (limit 2); kidney: 2.1 nTPM (limit 2); hypothalamus: 2.7 nTPM (baseline 2); kidney: 2.1 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (TCGA): 108/141 (76.6%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 177/283 (62.5%); Head and Neck Squamous Cell Carcinoma (TCGA): 248/492 (50.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcription factor involved in regulation of gene expression in a variety of processes, including formation of positional identity in the developing retina, regionalization of the optic chiasm, morphogenesis of the kidney, and neuralization of ectodermal cells (By similarity). Involved in transcriptional activation of PGF and C3 genes (PubMed:27805902)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q16676 | HPA portal (live; not the pinned values)

FOXG1 | Unsafe / RNA

ENSG00000176165

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 15 nTPM (limit 2); basal ganglia: 18.7 nTPM (limit 2); cerebral cortex: 20.6 nTPM (limit 2); hippocampal formation: 15.4 nTPM (limit 2); hypothalamus: 2.3 nTPM (limit 2); pituitary gland: 2.7 nTPM (limit 2); amygdala: 15 nTPM (baseline 2); basal ganglia: 18.7 nTPM (baseline 2); cerebral ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Glioblastoma Multiforme (TCGA): 128/141 (90.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 32/176 (18.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcription repression factor which plays an important role in the establishment of the regional subdivision of the developing brain and in the development of the telencephalon

Curated tissue note. Expression is restricted to the neurons of the developing telencephalon

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P55316 | HPA portal (live; not the pinned values)

FOXJ3 | Unsafe / RNA

ENSG00000198815

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 22.4 nTPM (limit 10); adrenal gland: 12.5 nTPM (limit 2); amygdala: 14.7 nTPM (limit 2); basal ganglia: 18 nTPM (limit 2); bone marrow: 29.8 nTPM (limit 2); cerebellum: 28.8 nTPM (limit 2); cerebral cortex: 18.5 nTPM (limit 2); choroid plexus: 12.1 nTPM (limit 2); colon: 17.2 nTPM ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcriptional activator of MEF2C involved in the regulation of adult muscle fiber type identity and skeletal muscle regeneration (By similarity). Plays an important role in spermatogenesis (By similarity). Required for the survival of spermatogonia and participates in spermatocyte meiosis (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UPW0 | HPA portal (live; not the pinned values)

FOXO3B | Unsafe / RNA

ENSG00000240445

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.2 nTPM (limit 2); cerebral cortex: 2.2 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Breast Invasive Carcinoma (validation): 47/50 (94.0%); Colon Adenocarcinoma (validation): 92/486 (18.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcription factor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0A2Z4LIS9 | HPA portal (live; not the pinned values)

FOXP3 | Unsafe / Protein

ENSG00000049768

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. lung / macrophages: Medium (Enhanced); lymph node / non-germinal center cells: Medium (Enhanced); spleen / cells in white pulp: Medium (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 36/50 (72.0%); Rectum Adenocarcinoma (validation): 138/207 (66.7%); Colon Adenocarcinoma (validation): 310/486 (63.8%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Transcriptional regulator which is crucial for the development and inhibitory function of regulatory T-cells (Treg) (PubMed:17377532, PubMed:21458306, PubMed:23947341, PubMed:24354325, PubMed:24722479, PubMed:24835996, PubMed:30513302, PubMed:32644293). Plays an essential role in maintaining homeostasis of the immune system by allowing the acquisition of full suppressive function and stability of the Treg lineage, and by directly ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BZS1 | HPA portal (live; not the pinned values)

FRMPD1 | Unsafe / Protein

ENSG00000070601

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); cerebellum / cells in granular layer: High (Uncertain); cerebellum / cells in ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 55/80 (68.8%); Kidney Chromophobe (TCGA): 13/64 (20.3%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 45/283 (15.9%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); stomach cancer: 9/9 (100.0%)

Biology. Stabilizes membrane-bound GPSM1, and thereby promotes its interaction with GNAI1

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5SYB0 | HPA portal (live; not the pinned values)

FSHR | Unsafe / High RNA

ENSG00000170820

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 67.6 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 0.08981 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 2/282 (0.7%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. G protein-coupled receptor for follitropin, the follicle-stimulating hormone (PubMed:11847099, PubMed:24058690, PubMed:24692546). Mediates FSH signaling through cAMP/protein kinase A (PKA) and phosphatidylinositol-3 kinase (PI3K)/protein kinase B (AKT) pathways to regulate ovarian follicular development and steroidogenesis (PubMed:24058690). Has diverse extragonadal functions: signals via a GNAI2-coupled FSHR on osteoclasts and their ... [full text in table]

Curated tissue note. Sertoli cells and ovarian granulosa cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P23945 | HPA portal (live; not the pinned values)

FSIP1 | Unsafe / RNA

ENSG00000150667

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. colon: 7.1 nTPM (limit 6); rectum: 10.6 nTPM (limit 6); colon: 7.1 nTPM (baseline 6); rectum: 10.6 nTPM (baseline 6)

Cancer RNA. Glioblastoma Multiforme (validation): 54/58 (93.1%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Breast Invasive Carcinoma (TCGA): 482/1022 (47.2%)

Cancer IHC. breast cancer: 12/12 (100.0%); endometrial cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NA03 | HPA portal (live; not the pinned values)

FSIP2 | Unsafe / High RNA

ENSG00000188738

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 71.8 nTPM; colon/distal enterocytes; hpa_single_cell_tissue; protected; 3.409 TPM; blood/CD8_NAIVE; dice_cd8_naive; vital

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Ovary Serous Cystadenocarcinoma (validation): 58/81 (71.6%); Colon Adenocarcinoma (validation): 21/486 (4.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays a role in spermatogenesis

Curated tissue note. Predominantly expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5CZC0 | HPA portal (live; not the pinned values)

GABPA | Unsafe / Protein

ENSG00000154727

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Supported); adrenal gland / glandular cells: Medium (Supported); bronchus / respiratory epithelial cells: Medium (Supported); cerebral cortex / endothelial cells: Low (Supported); cerebral cortex / glial cells: Low (Supported); cerebral cortex / neuropil: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. lymphoma: 10/12 (83.3%); breast cancer: 5/11 (45.5%)

Biology. Transcription factor capable of interacting with purine rich repeats (GA repeats). Positively regulates transcription of transcriptional repressor RHIT/ZNF205 (PubMed:22306510) (Microbial infection) Necessary for the expression of the Adenovirus E4 gene

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q06546 | HPA portal (live; not the pinned values)

GABRB3 | Unsafe / RNA

ENSG00000166206

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 5.2 nTPM (limit 2); amygdala: 3.2 nTPM (limit 2); basal ganglia: 9.4 nTPM (limit 2); cerebellum: 9.8 nTPM (limit 2); cerebral cortex: 45.7 nTPM (limit 2); heart muscle: 5.1 nTPM (limit 2); hippocampal formation: 4 nTPM (limit 2); hypothalamus: 5.9 nTPM (limit 2); kidney: 7 nTPM ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 332/480 (69.2%); Glioblastoma Multiforme (validation): 36/58 (62.1%); Testicular Germ Cell Tumor (TCGA): 76/133 (57.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Beta subunit of the heteropentameric ligand-gated chloride channel gated by gamma-aminobutyric acid (GABA), a major inhibitory neurotransmitter in the brain (PubMed:14993607, PubMed:18514161, PubMed:22243422, PubMed:22303015, PubMed:24909990, PubMed:26950270, PubMed:30602789). GABA-gated chloride channels, also named GABA(A) receptors (GABAAR), consist of five subunits arranged around a central pore and contain GABA active binding ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P28472 | HPA portal (live; not the pinned values)

GABRG3 | Unsafe / Protein

ENSG00000182256

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / processes in white matter: Low (Approved); cerebellum / Purkinje cells - cytoplasm/membrane: Low (Approved); cerebral cortex / neuronal cells: Medium (Approved); hippocampus / neuronal cells: ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 310/480 (64.6%); Thyroid Carcinoma (TCGA): 96/495 (19.4%); Ovary Serous Cystadenocarcinoma (TCGA): 39/349 (11.2%)

Cancer IHC. prostate cancer: 2/10 (20.0%); endometrial cancer: 1/12 (8.3%)

Biology. Gamma subunit of the heteropentameric ligand-gated chloride channel gated by gamma-aminobutyric acid (GABA), a major inhibitory neurotransmitter in the brain (By similarity). GABA-gated chloride channels, also named GABA(A) receptors (GABAAR), consist of five subunits arranged around a central pore and contain GABA active binding site(s) located at the alpha and beta subunit interface(s) (By similarity). When activated by GABA, GABAARs ... [full text in table]

Curated tissue note. Expressed in brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q99928 | HPA portal (live; not the pinned values)

GABRR1 | Unsafe / RNA

ENSG00000146276

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 23.9 nTPM (limit 2); retina: 23.9 nTPM (baseline 2)

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 32/489 (6.5%); Lung Squamous Cell Carcinoma (validation): 4/68 (5.9%); Testicular Germ Cell Tumor (TCGA): 6/133 (4.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Rho subunit of the pentameric ligand-gated chloride channels responsible for mediating the effects of gamma-aminobutyric acid (GABA), the major inhibitory neurotransmitter in the brain (PubMed:37659407). Rho-containing GABA-gated chloride channels are a subclass of GABA(A) receptors (GABAARs) entirely composed of rho subunits, where GABA molecules bind at the rho intersubunit interfaces (PubMed:37659407). When activated by GABA, ... [full text in table]

Curated tissue note. Highly expressed in the retina (PubMed:1849271). Expressed in a lesser extent in brain, lung and thymus (PubMed:1849271)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P24046 | HPA portal (live; not the pinned values)

GABRR3 | Unsafe / RNA

ENSG00000183185

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 8.5 nTPM (limit 2); retina: 8.5 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Liver Hepatocellular Carcinoma (validation): 1/231 (0.4%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Rho subunit of the pentameric ligand-gated chloride channels responsible for mediating the effects of gamma-aminobutyric acid (GABA), the major inhibitory neurotransmitter in the brain (By similarity). Rho-containing GABA-gated chloride channels are a subclass of GABA(A) receptors (GABAARs) entirely composed of rho subunits, where GABA molecules bind at the rho intersubunit interfaces (By similarity). When activated by GABA, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A8MPY1 | HPA portal (live; not the pinned values)

GAGE12B | Unsafe / RNA

ENSG00000236737

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 3 nTPM (limit 2); basal ganglia: 3 nTPM (baseline 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 18/349 (5.2%); Stomach Adenocarcinoma (TCGA): 16/346 (4.6%); Thyroid Carcinoma (TCGA): 15/495 (3.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

HPA portal (live; not the pinned values)

GAGE12F | Unsafe / High RNA

ENSG00000236362

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 83.07 nTPM; blood/T-reg; hpa_immune_cell_samples; vital; 71.67 TPM; blood/T-reg; hpa_immune_cell_samples; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 32/133 (24.1%); Lung Squamous Cell Carcinoma (TCGA): 115/489 (23.5%); Lung Squamous Cell Carcinoma (validation): 13/68 (19.1%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Expressed in some prostate cancer tissues but not in normal prostate tissue

Reconsideration. Review uniquely assigning reads and exact gene/protein identity before reconsidering local expression.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O76087 | UniProt P0CL82 | UniProt P0CL80 | UniProt P0CL81 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

GAGE12G | Unsafe / High RNA

ENSG00000215269

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 66.05 nTPM; blood/T-reg; hpa_immune_cell_samples; vital; 56.98 TPM; blood/T-reg; hpa_immune_cell_samples; vital

Cancer RNA. Ovary Serous Cystadenocarcinoma (validation): 6/81 (7.4%); Skin Cuteneous Melanoma (TCGA): 5/99 (5.1%); Ovary Serous Cystadenocarcinoma (TCGA): 16/349 (4.6%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Expressed in some prostate cancer tissues but not in normal prostate tissue

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O76087 | UniProt P0CL82 | UniProt P0CL80 | UniProt P0CL81 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

GAGE12J | Unsafe / RNA

ENSG00000224659

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. retina: 2.2 nTPM (limit 2)

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 10/68 (14.7%); Lung Squamous Cell Carcinoma (TCGA): 65/489 (13.3%); Skin Cuteneous Melanoma (TCGA): 10/99 (10.1%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NER3 | HPA portal (live; not the pinned values)

GAGE2E | Unsafe / RNA

ENSG00000275113

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.4 nTPM (limit 2); stomach: 14.9 nTPM (limit 8)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 28/133 (21.1%); Lung Squamous Cell Carcinoma (TCGA): 102/489 (20.9%); Skin Cuteneous Melanoma (TCGA): 17/99 (17.2%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4V326 | HPA portal (live; not the pinned values)

GALNTL5 | Unsafe / Protein

ENSG00000106648

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / neuronal cells: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Probable inactive glycosyltransferase required during spermatid development. May participate in protein loading into the acrosomes and accumulation of ubiquitin-proteasome systems around the head-tail coupling apparatus region

Curated tissue note. Mainly expressed in testis. Weakly or not expressed in other tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z4T8 | HPA portal (live; not the pinned values)

GAPDHS | Unsafe / RNA

ENSG00000105679

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 3.4 nTPM (limit 2)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 89/99 (89.9%); Rectum Adenocarcinoma (validation): 172/207 (83.1%); Colon Adenocarcinoma (validation): 316/486 (65.0%)

Cancer IHC. melanoma: 7/12 (58.3%); prostate cancer: 1/12 (8.3%)

Biology. Catalyzes the conversion of D-glyceraldehyde 3-phosphate (G3P) into 3-phospho-D-glyceroyl phosphate in glycolysis and the reverse reaction in gluconeogenesis (By similarity). May play an important role in regulating the switch between different pathways for energy production during spermiogenesis and in the spermatozoon. Required for sperm motility and male fertility (By similarity)

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14556 | HPA portal (live; not the pinned values)

GARIN1A | Unsafe / RNA

ENSG00000205085

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.2 nTPM (limit 2); pituitary gland: 4.3 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (validation): 48/58 (82.8%); Breast Invasive Carcinoma (validation): 20/50 (40.0%); Pancreatic Adenocarcinoma (validation): 15/80 (18.8%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. RAB2B effector protein required for accurate acrosome formation and normal male fertility

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6NXP2 | HPA portal (live; not the pinned values)

GARIN4 | Unsafe / High RNA

ENSG00000162771

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 39 nTPM; salivary gland/serous glandular cells; hpa_single_cell_tissue; protected; 1.017 TPM; blood/MONOCYTES; dice_monocytes; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. RAB2B effector protein required for the compacted Golgi morphology, probably through interaction with small GTPase RAB2B

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYT1 | HPA portal (live; not the pinned values)

GARIN5A | Unsafe / RNA

ENSG00000142530

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 7.4 nTPM (limit 2); basal ganglia: 9 nTPM (limit 2); cerebellum: 4.3 nTPM (limit 2); cerebral cortex: 7.9 nTPM (limit 2); choroid plexus: 2.6 nTPM (limit 2); hippocampal formation: 5.2 nTPM (limit 2); hypothalamus: 8.4 nTPM (limit 2); kidney: 3.4 nTPM (limit 2); liver: 2.6 nTPM (limit 2); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 46/50 (92.0%); Ovary Serous Cystadenocarcinoma (TCGA): 320/349 (91.7%); Prostate Adenocarcinoma (TCGA): 386/480 (80.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. RAB2B effector protein which promotes cytosolic DNA-induced innate immune responses. Regulates IFN responses against DNA viruses by regulating the CGAS-STING signaling axis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6IPT2 | HPA portal (live; not the pinned values)

GART | Unsafe / Protein

ENSG00000159131

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); melanoma: 11/11 (100.0%)

Biology. Trifunctional enzyme that catalyzes three distinct reactions as part of the 'de novo' inosine monophosphate biosynthetic pathway

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P22102 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

GAST | Unsafe / Protein

ENSG00000184502

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. stomach 2 / glandular cells: Medium (Enhanced)

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 211/492 (42.9%); Lung Squamous Cell Carcinoma (TCGA): 138/489 (28.2%); Lung Squamous Cell Carcinoma (validation): 13/68 (19.1%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Gastrin stimulates the stomach mucosa to produce and secrete hydrochloric acid and the pancreas to secrete its digestive enzymes. It also stimulates smooth muscle contraction and increases blood circulation and water secretion in the stomach and intestine

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P01350 | HPA portal (live; not the pinned values)

GATC | Unsafe / RNA

ENSG00000257218

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 10 nTPM (limit 2); amygdala: 9.9 nTPM (limit 2); basal ganglia: 9.1 nTPM (limit 2); bone marrow: 7.3 nTPM (limit 2); cerebellum: 15 nTPM (limit 2); cerebral cortex: 11.3 nTPM (limit 2); choroid plexus: 8.1 nTPM (limit 2); colon: 9.8 nTPM (limit 6); duodenum: 11.4 nTPM (limit 2); ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Structural subunit of the mitochondrial Glu-tRNA(Gln) amidotransferase complex GatCAB. GatCAB allows the formation of correctly charged Gln-tRNA(Gln) through the transamidation of misacylated Glu-tRNA(Gln) in the mitochondria (PubMed:19805282). The reaction takes place in the presence of glutamine and ATP through an activated gamma-phospho-Glu-tRNA(Gln) (By similarity). This subunit probably forms a belt that wraps around the interface ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43716 | HPA portal (live; not the pinned values)

GBX1 | Unsafe / RNA

ENSG00000164900

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skeletal muscle: 10.9 nTPM (limit 4)

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 30/283 (10.6%); Head and Neck Squamous Cell Carcinoma (TCGA): 10/492 (2.0%); Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14549 | HPA portal (live; not the pinned values)

GCM1 | Unsafe / High RNA

ENSG00000137270

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 48.1 nTPM; kidney/proximal tubular cells; hpa_single_cell_tissue; vital; 10.96 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 7/50 (14.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 16/282 (5.7%); Testicular Germ Cell Tumor (TCGA): 6/133 (4.5%)

Cancer IHC. lymphoma: 2/12 (16.7%); breast cancer: 0/12 (0.0%)

Biology. Transcription factor involved in the control of expression of placental growth factor (PGF) and other placenta-specific genes (PubMed:10542267, PubMed:18160678). Binds to the trophoblast-specific element 2 (TSE2) of the aromatase gene enhancer (PubMed:10542267). Binds to the SYDE1 promoter (PubMed:27917469). Has a central role in mediating the differentiation of trophoblast cells along both the villous and extravillous pathways in ... [full text in table]

Curated tissue note. Highly expressed in the placenta (PubMed:10542267). Expressed in trophoblast cells of the villi (PubMed:27917469)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NP62 | HPA portal (live; not the pinned values)

GDAP2 | Unsafe / Protein

ENSG00000196505

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: High (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. breast cancer: 8/9 (88.9%); urothelial cancer: 8/11 (72.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NXN4 | HPA portal (live; not the pinned values)

GDF5 | Unsafe / RNA

ENSG00000125965

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. lung: 2.2 nTPM (limit 2); salivary gland: 9.6 nTPM (limit 6); lung: 2.2 nTPM (baseline 2); salivary gland: 9.6 nTPM (baseline 6)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 55/176 (31.2%); Breast Invasive Carcinoma (validation): 5/50 (10.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 19/282 (6.7%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); lung cancer: 10/10 (100.0%)

Biology. Growth factor involved in bone and cartilage formation. During cartilage development regulates differentiation of chondrogenic tissue through two pathways. Firstly, positively regulates differentiation of chondrogenic tissue through its binding of high affinity with BMPR1B and of less affinity with BMPR1A, leading to induction of SMAD1-SMAD5-SMAD8 complex phosphorylation and then SMAD protein signaling transduction (PubMed:15530414, ... [full text in table]

Curated tissue note. Predominantly expressed in long bones during embryonic development. Expressed in monocytes (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P43026 | HPA portal (live; not the pinned values)

GDF6 | Unsafe / RNA

ENSG00000156466

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. heart muscle: 2.7 nTPM (limit 2); heart muscle: 2.7 nTPM (baseline 2)

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 47/100 (47.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 227/521 (43.6%); Testicular Germ Cell Tumor (TCGA): 11/133 (8.3%)

Cancer IHC. melanoma: 11/12 (91.7%); prostate cancer: 3/12 (25.0%)

Biology. Growth factor that controls proliferation and cellular differentiation in the retina and bone formation. Plays a key role in regulating apoptosis during retinal development. Establishes dorsal-ventral positional information in the retina and controls the formation of the retinotectal map (PubMed:23307924). Required for normal formation of bones and joints in the limbs, skull, digits and axial skeleton. Plays a key role in establishing ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6KF10 | HPA portal (live; not the pinned values)

GDPD1 | Unsafe / RNA

ENSG00000153982

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 5.2 nTPM (limit 2); basal ganglia: 5.8 nTPM (limit 2); cerebellum: 10 nTPM (limit 2); cerebral cortex: 11.6 nTPM (limit 2); choroid plexus: 2.9 nTPM (limit 2); duodenum: 5.6 nTPM (limit 2); hippocampal formation: 6 nTPM (limit 2); hypothalamus: 7.6 nTPM (limit 2); lung: 2.2 nTPM (limit ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Glioblastoma Multiforme (TCGA): 95/141 (67.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Hydrolyzes lysoglycerophospholipids to produce lysophosphatidic acid (LPA) and the corresponding amines (PubMed:25596343, PubMed:27637550). Shows a preference for 1-O-alkyl-sn-glycero-3-phosphocholine (lyso-PAF), lysophosphatidylethanolamine (lyso-PE) and lysophosphatidylcholine (lyso-PC) (PubMed:25596343, PubMed:27637550). May be involved in bioactive N-acylethanolamine biosynthesis from both N-acyl-lysoplasmenylethanolamin ... [full text in table]

Curated tissue note. Widely expressed with high expression level in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N9F7 | HPA portal (live; not the pinned values)

GEN1 | Unsafe / Protein

ENSG00000178295

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); cerebellum / cells in granular layer: Medium (Enhanced); cerebellum / Purkinje cells: Medium (Enhanced); cerebral cortex / glial ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 483/486 (99.4%); Rectum Adenocarcinoma (validation): 204/207 (98.6%)

Cancer IHC. pancreatic cancer: 6/7 (85.7%); liver cancer: 9/12 (75.0%)

Biology. Endonuclease which resolves Holliday junctions (HJs) by the introduction of symmetrically related cuts across the junction point, to produce nicked duplex products in which the nicks can be readily ligated. Four-way DNA intermediates, also known as Holliday junctions, are formed during homologous recombination and DNA repair, and their resolution is necessary for proper chromosome segregation (PubMed:19020614, PubMed:26682650). Cleaves ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q17RS7 | HPA portal (live; not the pinned values)

GGN | Unsafe / RNA

ENSG00000179168

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.1 nTPM (limit 2); duodenum: 2.7 nTPM (limit 2); small intestine: 2.8 nTPM (limit 2); cerebral cortex: 2.1 nTPM (baseline 2); duodenum: 2.7 nTPM (baseline 2); small intestine: 2.8 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 6/50 (12.0%); Glioblastoma Multiforme (TCGA): 8/141 (5.7%); Ovary Serous Cystadenocarcinoma (TCGA): 17/349 (4.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in spermatogenesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86UU5 | HPA portal (live; not the pinned values)

GGTLC2 | Unsafe / RNA

ENSG00000100121

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. bone marrow: 2.2 nTPM (limit 2); bone marrow: 2.2 nTPM (baseline 2)

Cancer RNA. Kidney Renal Papillary Cell Carcinoma (TCGA): 24/282 (8.5%); Thyroid Carcinoma (TCGA): 36/495 (7.3%); Lung Adenocarcinoma (TCGA): 20/497 (4.0%)

Cancer IHC. renal cancer: 5/12 (41.7%); liver cancer: 4/11 (36.4%)

Biology. Placenta and sigmoid tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14390 | HPA portal (live; not the pinned values)

GH2 | Unsafe / Protein

ENSG00000136487

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. pituitary gland / cells in anterior : High (Supported)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%); Uterine Corpus Endometrial Carcinoma (TCGA): 2/176 (1.1%); Lung Adenocarcinoma (TCGA): 3/497 (0.6%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Plays an important role in growth control. Its major role in stimulating body growth is to stimulate the liver and other tissues to secrete IGF1. It stimulates both the differentiation and proliferation of myoblasts. It also stimulates amino acid uptake and protein synthesis in muscle and other tissues

Curated tissue note. Expressed in the placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P01242 | HPA portal (live; not the pinned values)

GINS1 | Unsafe / Protein

ENSG00000101003

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / Purkinje cells: Low (Uncertain); cerebral cortex / neuronal cells: Low ... [full text in table]

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 280/283 (98.9%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Colon Adenocarcinoma (validation): 473/486 (97.3%)

Cancer IHC. renal cancer: 11/12 (91.7%); testis cancer: 8/9 (88.9%)

Biology. Required for correct functioning of the GINS complex, a complex that plays an essential role in the initiation of DNA replication, and progression of DNA replication forks (PubMed:17417653, PubMed:28414293). GINS complex is a core component of CDC45-MCM-GINS (CMG) helicase, the molecular machine that unwinds template DNA during replication, and around which the replisome is built (PubMed:32453425, PubMed:34694004, PubMed:34700328, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14691 | HPA portal (live; not the pinned values)

GINS4 | Unsafe / Protein

ENSG00000147536

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 441/486 (90.7%); Testicular Germ Cell Tumor (TCGA): 120/133 (90.2%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 248/283 (87.6%)

Cancer IHC. melanoma: 9/9 (100.0%); prostate cancer: 10/11 (90.9%)

Biology. Required for correct functioning of the GINS complex, a complex that plays an essential role in the initiation of DNA replication, and progression of DNA replication forks (PubMed:17417653, PubMed:28414293). GINS complex is a core component of CDC45-MCM-GINS (CMG) helicase, the molecular machine that unwinds template DNA during replication, and around which the replisome is built (PubMed:32453425, PubMed:34694004, PubMed:34700328, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BRT9 | HPA portal (live; not the pinned values)

GIP | Unsafe / Protein

ENSG00000159224

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. duodenum / endocrine cells: High (Enhanced); small intestine / endocrine cells: High (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 94/133 (70.7%); Pancreatic Adenocarcinoma (TCGA): 6/176 (3.4%); Stomach Adenocarcinoma (TCGA): 10/346 (2.9%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Potent stimulator of insulin secretion and relatively poor inhibitor of gastric acid secretion

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P09681 | HPA portal (live; not the pinned values)

GJA8 | Unsafe / Protein

ENSG00000121634

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. eye / lens fiber cells: Medium (Enhanced)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 7/349 (2.0%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 2/282 (0.7%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Structural component of eye lens gap junctions (PubMed:18006672, PubMed:19756179). Gap junctions are dodecameric channels that connect the cytoplasm of adjoining cells. They are formed by the docking of two hexameric hemichannels, one from each cell membrane (By similarity). Small molecules and ions diffuse from one cell to a neighboring cell via the central pore (PubMed:18006672, PubMed:19756179)

Curated tissue note. Eye lens

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P48165 | HPA portal (live; not the pinned values)

GKAP1 | Unsafe / Protein

ENSG00000165113

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebellum / Purkinje cells - cytoplasm/membrane: Low (Enhanced); cerebellum / Purkinje cells - dendrites: Low (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Prostate Adenocarcinoma (TCGA): 447/480 (93.1%); Breast Invasive Carcinoma (validation): 46/50 (92.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Regulates insulin-dependent IRS1 tyrosine phosphorylation in adipocytes by modulating the availability of IRS1 to IR tyrosine kinase. Its association with IRS1 is required for insulin-induced translocation of SLC2A4 to the cell membrane. Involved in TNF-induced impairment of insulin-dependent IRS1 tyrosine phosphorylation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VSY0 | HPA portal (live; not the pinned values)

GLIPR1L2 | Unsafe / Protein

ENSG00000180481

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); lymph node / non-germinal center cells: Low (Enhanced)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 74/80 (92.5%); Glioblastoma Multiforme (validation): 29/58 (50.0%); Breast Invasive Carcinoma (validation): 19/50 (38.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Highly expressed in testis. Detected in prostate, kidney, bladder, lung and bone marrow

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4G1C9 | HPA portal (live; not the pinned values)

GLUD2 | Unsafe / Protein

ENSG00000182890

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: High (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 46/50 (92.0%); Glioblastoma Multiforme (validation): 29/58 (50.0%); Liver Hepatocellular Carcinoma (validation): 89/231 (38.5%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Catalyzes the oxidative deamination of glutamate to alpha-ketoglutarate and ammonia (PubMed:11032875). Plays a key role in glutamate metabolism and energy production via the TCA cycle in neural and testicular tissues (PubMed:11032875). Important for recycling glutamate during neurotransmission (By similarity)

Curated tissue note. Expressed in retina, testis and, at a lower level, brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P49448 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

GMCL1 | Unsafe / Protein

ENSG00000087338

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: High (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / goblet cells: High (Approved); caudate / ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. lung cancer: 8/10 (80.0%); melanoma: 9/12 (75.0%)

Biology. Possible function in spermatogenesis. Enhances the degradation of MDM2 and increases the amount of p53 probably by modulating the nucleocytoplasmic transport (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96IK5 | HPA portal (live; not the pinned values)

GMCL2 | Unsafe / Protein

ENSG00000244234

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / basal cells: High (Uncertain); bronchus / ciliated cells (cell body): High (Uncertain); bronchus / goblet cells: High (Uncertain); ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (TCGA): 2/362 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 1/282 (0.4%)

Cancer IHC. lung cancer: 8/10 (80.0%); melanoma: 9/12 (75.0%)

Biology. Possible function in spermatogenesis. Probable substrate-specific adapter of an E3 ubiquitin-protein ligase complex which mediates the ubiquitination and subsequent proteasomal degradation of target proteins (PubMed:14528312)

Curated tissue note. Expressed predominantly in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEA9 | HPA portal (live; not the pinned values)

GML | Unsafe / RNA

ENSG00000104499

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 187.3 nTPM (limit 2); adrenal gland: 187.3 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Liver Hepatocellular Carcinoma (TCGA): 1/362 (0.3%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May play a role in the apoptotic pathway or cell-cycle regulation induced by p53/TP53 after DNA damage

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q99445 | HPA portal (live; not the pinned values)

GMNC | Unsafe / RNA

ENSG00000205835

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.9 nTPM (limit 2); liver: 4 nTPM (limit 2); choroid plexus: 2.9 nTPM (baseline 2); liver: 4 nTPM (baseline 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 102/349 (29.2%); Ovary Serous Cystadenocarcinoma (validation): 21/81 (25.9%); Glioblastoma Multiforme (validation): 7/58 (12.1%)

Cancer IHC. cervical cancer: 4/9 (44.4%); pancreatic cancer: 2/11 (18.2%)

Biology. Regulator of DNA replication. Promotes initiation of chromosomal DNA replication by mediating TOPBP1- and CDK2-dependent recruitment of CDC45L onto replication origins (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NCL1 | HPA portal (live; not the pinned values)

GNAL | Unsafe / Protein

ENSG00000141404

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bronchus / ciliated cells (cell body): Low (Enhanced); caudate / neuronal cells: Low (Enhanced); caudate / synapses: High (Enhanced); cerebellum / granular cells - cytoplasm/membrane: Low (Enhanced); cerebellum / synaptic glomeruli - core: Medium ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 285/480 (59.4%); Rectum Adenocarcinoma (validation): 121/207 (58.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 154/282 (54.6%)

Cancer IHC. pancreatic cancer: 3/10 (30.0%); cervical cancer: 2/12 (16.7%)

Biology. Guanine nucleotide-binding protein (G protein) involved as transducer in olfactory signal transduction controlled by G protein-coupled receptors (GPCRs) (By similarity). Contains the guanine nucleotide binding site and alternates between an active, GTP-bound state and an inactive, GDP-bound state (By similarity). Signaling by an activated GPCR promotes GDP release and GTP binding (By similarity). The alpha subunit has a low GTPase ... [full text in table]

Curated tissue note. Detected in olfactory neuroepithelium, brain, testis, and to a lower extent in retina, lung alveoli, spleen (PubMed:8243272). Trace amounts where seen in kidney, adrenal gland and liver (PubMed:8243272). Found ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P38405 | HPA portal (live; not the pinned values)

GNAT1 | Unsafe / Protein

ENSG00000114349

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: High (Enhanced)

Cancer RNA. Kidney Chromophobe (TCGA): 1/64 (1.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 1/282 (0.4%)

Cancer IHC. testis cancer: 4/12 (33.3%); lung cancer: 2/11 (18.2%)

Biology. Alpha subunit of the heterotrimeric guanine nucleotide-binding protein (G protein) that transduces signals from the G protein-coupled photoreceptor rhodopsin (RHO). Essential for normal RHO-mediated light perception in retinal rod photoreceptors (PubMed:22190596). Functions as a GTPase, cycling between an active GTP-bound state and an inactive GDP-bound state. Under dim light conditions, photoactivated RHO promotes GDP release and GTP ... [full text in table]

Curated tissue note. Rod photoreceptor cells (PubMed:1614872). Predominantly expressed in the retina followed by the ciliary body, iris and retinal pigment epithelium (PubMed:22190596)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P11488 | HPA portal (live; not the pinned values)

GNAT3 | Unsafe / Protein

ENSG00000214415

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. duodenum / endocrine cells: High (Enhanced); small intestine / endocrine cells: High (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 2/50 (4.0%); Bladder Urothelial Carcinoma (TCGA): 6/169 (3.6%); Pancreatic Adenocarcinoma (validation): 2/80 (2.5%)

Cancer IHC. testis cancer: 1/12 (8.3%); breast cancer: 0/9 (0.0%)

Biology. Guanine nucleotide-binding protein (G protein) alpha subunit playing a prominent role in bitter and sweet taste transduction as well as in umami (monosodium glutamate, monopotassium glutamate, and inosine monophosphate) taste transduction (PubMed:40339580, PubMed:38600377, PubMed:38776963). Transduction by this alpha subunit involves coupling of specific cell-surface receptors with a cGMP-phosphodiesterase; Activation of ... [full text in table]

Curated tissue note. Expressed in taste buds (sensory organs of clustered epithelial cells) of the circumvallate and foliate papillae of the tongue at protein level. Expressed in enteroendocrine L cells of the gut. Detected also ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A8MTJ3 | HPA portal (live; not the pinned values)

GNG2 | Unsafe / Protein

ENSG00000186469

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebellum / cells in molecular layer: Low (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. melanoma: 7/12 (58.3%); breast cancer: 4/11 (36.4%)

Biology. Guanine nucleotide-binding proteins (G proteins) are involved as a modulator or transducer in various transmembrane signaling systems (PubMed:29925951, PubMed:33762731, PubMed:34239069, PubMed:35610220, PubMed:35714614, PubMed:35835867, PubMed:36087581, PubMed:36989299, PubMed:37327704, PubMed:37935376, PubMed:37935377, PubMed:37963465, PubMed:38168118, PubMed:38552625, PubMed:39103320). The beta and gamma chains are required for the ... [full text in table]

Curated tissue note. Expressed in fetal tissues, including testis, adrenal gland, brain, white blood cells and brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P59768 | HPA portal (live; not the pinned values)

GNGT1 | Unsafe / Protein

ENSG00000127928

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: High (Enhanced)

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 21/100 (21.0%); Kidney Chromophobe (TCGA): 9/64 (14.1%); Breast Invasive Carcinoma (validation): 5/50 (10.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Guanine nucleotide-binding proteins (G proteins) are involved as a modulator or transducer in various transmembrane signaling systems. The beta and gamma chains are required for the GTPase activity, for replacement of GDP by GTP, and for G protein-effector interaction

Curated tissue note. Retinal rod outer segment

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P63211 | HPA portal (live; not the pinned values)

GOPC | Unsafe / Protein

ENSG00000047932

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: Low (Approved); bronchus / ciliated cells (cell body): Medium (Approved); bronchus / ciliated cells (tip of cilia): Low (Approved); bronchus / goblet cells: Medium ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. glioma: 2/11 (18.2%); liver cancer: 2/12 (16.7%)

Biology. Plays a role in intracellular protein trafficking and degradation (PubMed:11707463, PubMed:14570915, PubMed:15358775). May regulate CFTR chloride currents and acid-induced ASIC3 currents by modulating cell surface expression of both channels (By similarity). May also regulate the intracellular trafficking of the ADR1B receptor (PubMed:15358775). May play a role in autophagy (By similarity). Together with MARCHF2 mediates the ... [full text in table]

Curated tissue note. Ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HD26 | HPA portal (live; not the pinned values)

GP1BA | Unsafe / Protein

ENSG00000185245

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Supported); spleen / cells in red pulp: Medium (Supported)

Cancer RNA. Breast Invasive Carcinoma (validation): 11/50 (22.0%); Testicular Germ Cell Tumor (TCGA): 24/133 (18.0%); Rectum Adenocarcinoma (validation): 13/207 (6.3%)

Cancer IHC. ovarian cancer: 1/11 (9.1%); glioma: 1/12 (8.3%)

Biology. GP-Ib, a surface membrane protein of platelets, participates in the formation of platelet plugs by binding to the A1 domain of vWF, which is already bound to the subendothelium

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P07359 | HPA portal (live; not the pinned values)

GPD2 | Unsafe / Protein

ENSG00000115159

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Enhanced); adrenal gland / glandular cells: High (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: High (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / granular cells - ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); endometrial cancer: 12/12 (100.0%)

Biology. Calcium-responsive mitochondrial glycerol-3-phosphate dehydrogenase which seems to be a key component of the pancreatic beta-cell glucose-sensing device

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P43304 | HPA portal (live; not the pinned values)

GPR18 | Unsafe / Protein

ENSG00000125245

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. lymph node / germinal center cells: Low (Enhanced); lymph node / non-germinal center cells: Medium (Enhanced); spleen / cells in red pulp: Medium (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 27/50 (54.0%); Rectum Adenocarcinoma (validation): 77/207 (37.2%); Colon Adenocarcinoma (validation): 169/486 (34.8%)

Cancer IHC. lymphoma: 3/12 (25.0%); glioma: 1/11 (9.1%)

Biology. G protein-coupled receptor (GPCR) that plays a role in diverse physiological processes particularly within the immune and nervous systems (PubMed:21732409, PubMed:26195725). Becomes active when triggered by various endogenous ligands including endocannabinoid N-arachidonyl glycine (NAGly), delta-9-tetrahydrocannabinol or resolvin D2/RvD2 derived from the omega-3 fatty acid docosahexaenoic acid (DHA) (PubMed:16844083, PubMed:24762058, ... [full text in table]

Curated tissue note. Expressed in midpiece of spermatozoon (at protein level) (PubMed:27572937). Most abundant in testis and spleen (PubMed:16844083). Highly expressed in CD4 and CD8-positive T-cells as well as CD19-positive ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14330 | HPA portal (live; not the pinned values)

GPR32 | Unsafe / RNA

ENSG00000142511

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. parathyroid gland: 56.2 nTPM (limit 6); parathyroid gland: 56.2 nTPM (baseline 6)

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 2/169 (1.2%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. G protein-coupled receptor that binds to several ligands including resolvin D1 (RvD1) with high affinity, leading to rapid and transient activation of numerous intracellular signaling pathways. In macrophages, enhances the RvD1-stimulated phagocytic and clearance functions (PubMed:20080636). Macrophages migrate less toward different chemoattractant stimuli but phagocytose more microbial particles (PubMed:26969756). Prevents the ... [full text in table]

Curated tissue note. Expressed in resting primary human macrophages

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75388 | HPA portal (live; not the pinned values)

GPR39 | Unsafe / Protein

ENSG00000183840

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Low (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); cerebral cortex / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 199/207 (96.1%); Colon Adenocarcinoma (validation): 449/486 (92.4%); Lung Adenocarcinoma (TCGA): 444/497 (89.3%)

Cancer IHC. endometrial cancer: 2/11 (18.2%); colorectal cancer: 1/11 (9.1%)

Biology. Zinc-sensing receptor that can sense changes in extracellular Zn(2+), mediate Zn(2+) signal transmission, and participates in the regulation of numerous physiological processes including glucose homeostasis regulation, gastrointestinal mobility, hormone secretion and cell death (PubMed:18180304). Activation by Zn(2+) in keratinocytes increases the intracellular concentration of Ca(2+) and activates the ERK/MAPK and PI3K/AKT signaling ... [full text in table]

Curated tissue note. Expressed in many tissues, including the stomach, intestine and hypothalamus

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43194 | HPA portal (live; not the pinned values)

GPR55 | Unsafe / RNA

ENSG00000135898

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 3.5 nTPM (limit 2); small intestine: 2.1 nTPM (limit 2); basal ganglia: 3.5 nTPM (baseline 2); small intestine: 2.1 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 9/133 (6.8%); Rectum Adenocarcinoma (validation): 9/207 (4.3%); Breast Invasive Carcinoma (validation): 2/50 (4.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. G protein-coupled receptor that binds to several ligands including 2-arachidonoyl lysophosphatidylinositol or lysophosphatidylglucoside with high affinity, leading to rapid and transient activation of numerous intracellular signaling pathways (PubMed:36142844, PubMed:36523570, PubMed:37544935). Induces the Ca(2+) release from intracellular stores via ERK, the heterotrimeric G protein GNA13 and RHOA leading to morphological changes ... [full text in table]

Curated tissue note. Expressed in the caudate nucleus and putamen, but not detected in the hippocampus, thalamus, pons cerebellum, frontal cortex of the brain or in the liver. Expressed in osteoclasts and osteoblasts. Highly ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y2T6 | HPA portal (live; not the pinned values)

GPR89B | Unsafe / RNA

ENSG00000188092

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 6.8 nTPM (limit 2); amygdala: 5 nTPM (limit 2); basal ganglia: 5 nTPM (limit 2); bone marrow: 3 nTPM (limit 2); cerebellum: 6.8 nTPM (limit 2); cerebral cortex: 6.5 nTPM (limit 2); heart muscle: 5.5 nTPM (limit 2); hippocampal formation: 4.8 nTPM (limit 2); hypothalamus: 6.8 nTPM ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 79/80 (98.8%); Colon Adenocarcinoma (validation): 479/486 (98.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Voltage-gated channel that enables the transfer of monoatomic anions such as iodide, chloride, bromide and fluoride which may function in counter-ion conductance and participates in Golgi acidification (PubMed:18794847). Plays a role in lymphocyte development, probably by acting as a RABL3 effector in hematopoietic cells (By similarity). Essential for maintaining the acidic luminal pH of the Golgi apparatus in neurons, which is ... [full text in table]

Curated tissue note. Ubiquitous

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt B7ZAQ6 | UniProt P0CG08 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

GRID2 | Unsafe / Protein

ENSG00000152208

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebellum / processes in molecular layer: High (Enhanced); cerebellum / Purkinje cells - cytoplasm/membrane: Medium (Enhanced); cerebellum / synaptic glomeruli - core: Medium (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 43/58 (74.1%); Glioblastoma Multiforme (TCGA): 45/141 (31.9%); Testicular Germ Cell Tumor (TCGA): 6/133 (4.5%)

Cancer IHC. stomach cancer: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. Member of the ionotropic glutamate receptor family, which plays a crucial role in synaptic organization and signal transduction in the central nervous system (PubMed:34936451, PubMed:27418511, PubMed:40957579). Although it shares structural features with ionotropic glutamate receptors, does not bind glutamate as a primary ligand (PubMed:34936451). Promotes synaptogenesis and mediates the D-serine-dependent long term depression signals ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43424 | HPA portal (live; not the pinned values)

GRIK5 | Unsafe / RNA

ENSG00000105737

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.4 nTPM (limit 2); amygdala: 16.9 nTPM (limit 2); basal ganglia: 20.6 nTPM (limit 2); bone marrow: 2.7 nTPM (limit 2); cerebellum: 32.7 nTPM (limit 2); cerebral cortex: 27.3 nTPM (limit 2); choroid plexus: 7 nTPM (limit 2); colon: 15.9 nTPM (limit 6); heart muscle: 4.8 nTPM (limit ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Glioblastoma Multiforme (TCGA): 137/141 (97.2%); Thyroid Carcinoma (TCGA): 402/495 (81.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Ionotropic glutamate receptor that functions as a cation-permeable ligand-gated ion channel, gated by L-glutamate and the glutamatergic agonist kainic acid. Cannot form functional channels on its own and produces channel activity only in heteromeric assembly with GRIK1 and GRIK2 subunits (PubMed:1321949, PubMed:14511640, PubMed:8730589). Can form functional heteromeric receptors with GRIK3 (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q16478 | HPA portal (live; not the pinned values)

GRIN3A | Unsafe / RNA

ENSG00000198785

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 7.4 nTPM (limit 2); cerebral cortex: 7.4 nTPM (baseline 2)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 85/480 (17.7%); Glioblastoma Multiforme (validation): 3/58 (5.2%); Breast Invasive Carcinoma (validation): 1/50 (2.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of a non-conventional N-methyl-D-aspartate (NMDA) receptors (NMDARs) that function as heterotetrameric, ligand-gated cation channels with low calcium permeability and low voltage-dependent block by Mg(2+) (By similarity). During the development of neural circuits, participates in the synaptic refinement period, restricting spine maturation and growth (By similarity). Forms glutamatergic receptor complexes with GluN1 and GluN2 ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TCU5 | HPA portal (live; not the pinned values)

GRK4 | Unsafe / Protein

ENSG00000125388

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): High (Approved); cerebral cortex / neuronal cells: Low (Approved); nasopharynx / ciliated cells (ciliary rootlets): High (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Glioblastoma Multiforme (TCGA): 107/141 (75.9%)

Cancer IHC. liver cancer: 6/12 (50.0%); colorectal cancer: 2/12 (16.7%)

Biology. Specifically phosphorylates the activated forms of G protein-coupled receptors, such as ADRB2, DRD1 and DRD3 (PubMed:16338988, PubMed:19520868). Such receptor phosphorylation initiates beta-arrestin-mediated receptor internalization, and signaling events leading to their desensitization (PubMed:16338988, PubMed:19520868). Mediates phosphorylation of DRD1 in the absence of dopamine activation, resulting in constitutive desensitization ... [full text in table]

Curated tissue note. Isoform 1, isoform 2, isoform 3, and isoform 4 are expressed in testis. Isoform 4 is expressed in myometrium

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P32298 | HPA portal (live; not the pinned values)

GRM8 | Unsafe / RNA

ENSG00000179603

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.1 nTPM (limit 2); choroid plexus: 21 nTPM (limit 2); liver: 2.7 nTPM (limit 2); cerebral cortex: 2.1 nTPM (baseline 2); choroid plexus: 21 nTPM (baseline 2); liver: 2.7 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 155/207 (74.9%); Colon Adenocarcinoma (validation): 276/486 (56.8%); Rectum Adenocarcinoma (TCGA): 43/88 (48.9%)

Cancer IHC. breast cancer: 2/12 (16.7%); testis cancer: 2/12 (16.7%)

Biology. G protein-coupled receptor for glutamate. Ligand binding causes a conformation change that triggers signaling via guanine nucleotide-binding proteins (G proteins) and modulates the activity of down-stream effectors, such as adenylate cyclase. Signaling inhibits adenylate cyclase activity

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00222 | HPA portal (live; not the pinned values)

GRXCR1 | Unsafe / High RNA

ENSG00000215203

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 95.4 nTPM; prostate/t-cells; hpa_single_cell_tissue; vital; 0.3137 TPM; blood/TH2; dice_th2; vital

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. colorectal cancer: 8/11 (72.7%); breast cancer: 8/12 (66.7%)

Biology. May play a role in actin filament architecture in developing stereocilia of sensory cells

Curated tissue note. Expressed at low levels in adult lung, brain and duodenum with moderate levels in testis. Highly expressed in fetal cochlea

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A8MXD5 | HPA portal (live; not the pinned values)

GSG1 | Unsafe / RNA

ENSG00000111305

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 11.5 nTPM (limit 2); pancreas: 4.8 nTPM (limit 2); retina: 29.5 nTPM (limit 2); choroid plexus: 11.5 nTPM (baseline 2); pancreas: 4.8 nTPM (baseline 2); retina: 29.5 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 30/133 (22.6%); Kidney Renal Papillary Cell Carcinoma (TCGA): 44/282 (15.6%); Uterine Corpus Endometrial Carcinoma (TCGA): 16/176 (9.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May cause the redistribution of PAPOLB from the cytosol to the endoplasmic reticulum

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q2KHT4 | HPA portal (live; not the pinned values)

GSTA5 | Unsafe / High RNA

ENSG00000182793

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 102.9 nTPM; bronchus/smooth muscle cells; hpa_single_cell_tissue; vital; 0 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%); Breast Invasive Carcinoma (validation): 0/50 (0.0%)

Cancer IHC. liver cancer: 8/10 (80.0%); stomach cancer: 7/9 (77.8%)

Biology. Expression not detected

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7RTV2 | HPA portal (live; not the pinned values)

GSTCD | Unsafe / Protein

ENSG00000138780

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. colorectal cancer: 10/10 (100.0%); liver cancer: 10/12 (83.3%)

Biology. Widely expressed in cell types relevant to airway function, including airway smooth muscle cells and epithelial cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEC7 | HPA portal (live; not the pinned values)

GTF2A1L | Unsafe / RNA

ENSG00000242441

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. colon: 8.4 nTPM (limit 6); colon: 8.4 nTPM (baseline 6)

Cancer RNA. Breast Invasive Carcinoma (validation): 1/50 (2.0%); Glioblastoma Multiforme (validation): 1/58 (1.7%); Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May function as a testis specific transcription factor. Binds DNA in conjunction with GTF2A2 and TBP (the TATA-binding protein) and together with GTF2A2, allows mRNA transcription

Curated tissue note. Testis specific. Detected in adult testis mostly in round and elongating spermatids (at protein level). Detected in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UNN4 | HPA portal (live; not the pinned values)

GTPBP10 | Unsafe / Protein

ENSG00000105793

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: High (Approved); cerebellum / cells in molecular layer: Medium (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. skin cancer: 11/11 (100.0%); cervical cancer: 6/11 (54.5%)

Biology. May be involved in the ribosome maturation process. Complements an ObgE(CgtA) function in E.coli ribosome maturation. Plays a role of GTPase in vitro. When missing, disorganization of the nucleolar architecture is observed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A4D1E9 | HPA portal (live; not the pinned values)

GTSE1 | Unsafe / RNA

ENSG00000075218

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 11 nTPM (limit 2); duodenum: 4 nTPM (limit 2); lymph node: 9 nTPM (limit 6); small intestine: 2.8 nTPM (limit 2); thymus: 19.1 nTPM (limit 10); bone marrow: 11 nTPM (baseline 2); duodenum: 4 nTPM (baseline 2); lymph node: 9 nTPM (baseline 6); small intestine: 2.8 nTPM (baseline 2); ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 131/133 (98.5%); Colon Adenocarcinoma (validation): 473/486 (97.3%); Breast Invasive Carcinoma (validation): 47/50 (94.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in p53-induced cell cycle arrest in G2/M phase by interfering with microtubule rearrangements that are required to enter mitosis. Overexpression delays G2/M phase progression

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NYZ3 | HPA portal (live; not the pinned values)

GTSF1 | Unsafe / RNA

ENSG00000170627

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 6 nTPM (limit 4)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 102/133 (76.7%); Liver Hepatocellular Carcinoma (validation): 65/231 (28.1%); Lung Squamous Cell Carcinoma (validation): 18/68 (26.5%)

Cancer IHC. testis cancer: 7/11 (63.6%); lymphoma: 4/12 (33.3%)

Biology. Required for spermatogenesis and is involved in the suppression of retrotransposon transcription in male germ cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WW33 | HPA portal (live; not the pinned values)

GUCA1A | Unsafe / Protein

ENSG00000048545

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: High (Supported)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 80/133 (60.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 12/176 (6.8%); Bladder Urothelial Carcinoma (TCGA): 5/169 (3.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Stimulates retinal guanylyl cyclase when free calcium ions concentration is low and inhibits guanylyl cyclase when free calcium ions concentration is elevated (PubMed:18706439, PubMed:19459154, PubMed:30184081, PubMed:30622141). This Ca(2+)-sensitive regulation of retinal guanylyl cyclase is a key event in recovery of the dark state of rod photoreceptors following light exposure (By similarity). May be involved in cone photoreceptor ... [full text in table]

Curated tissue note. In the retina, it is expressed in rod and cone photoreceptors

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P43080 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

GUCY2F | Unsafe / RNA

ENSG00000101890

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 40.9 nTPM (limit 2); retina: 40.9 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Responsible for the synthesis of cyclic GMP (cGMP) in rods and cones of photoreceptors (PubMed:7777544). Plays an essential role in phototransduction, by mediating cGMP replenishment (By similarity). May also participate in the trafficking of membrane-associated proteins to the photoreceptor outer segment membrane (By similarity)

Curated tissue note. Retina. Localized exclusively in the outer nuclear layer and inner segments of the rod and cone photoreceptor cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P51841 | HPA portal (live; not the pinned values)

H1-1 | Unsafe / Protein

ENSG00000124610

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. esophagus / squamous epithelial cells: Low (Enhanced); kidney / distal tubules: Medium (Enhanced); nasopharynx / respiratory epithelial cells: Low (Enhanced); oral mucosa / squamous epithelial cells: Low (Enhanced); skin 1 / keratinocytes: Low (Enhanced); urinary bladder / urothelial cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 44/58 (75.9%); Pancreatic Adenocarcinoma (validation): 21/80 (26.2%); Colon Adenocarcinoma (validation): 97/486 (20.0%)

Cancer IHC. testis cancer: 3/11 (27.3%); endometrial cancer: 3/12 (25.0%)

Biology. Histone H1 protein binds to linker DNA between nucleosomes forming the macromolecular structure known as the chromatin fiber (PubMed:11130728). Histones H1 are necessary for the condensation of nucleosome chains into higher-order structured fibers and promote formation of the H3K27me3 mark by the PRC2/EED-EZH2 complex (PubMed:11130728). Also acts as a regulator of individual gene transcription through chromatin remodeling, nucleosome ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q02539 | HPA portal (live; not the pinned values)

H1-7 | Unsafe / Protein

ENSG00000187166

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); cerebellum / Purkinje cells: Low (Approved); colon / endothelial cells: Medium (Approved); duodenum / glandular cells: Low (Approved); ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 1/283 (0.4%)

Cancer IHC. testis cancer: 7/11 (63.6%); urothelial cancer: 4/9 (44.4%)

Biology. Essential for normal spermatogenesis and male fertility (PubMed:16533358). Required for proper cell restructuring and DNA condensation during the elongation phase of spermiogenesis. Involved in the histone-protamine transition of sperm chromatin and the subsequent production of functional sperm. Binds both double-stranded and single-stranded DNA, ATP and protamine-1

Curated tissue note. Testis-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q75WM6 | HPA portal (live; not the pinned values)

H1-8 | Unsafe / Protein

ENSG00000178804

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebellum / cells in molecular layer: Low (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May play a key role in the control of gene expression during oogenesis and early embryogenesis, presumably through the perturbation of chromatin structure. Essential for meiotic maturation of germinal vesicle-stage oocytes. The somatic type linker histone H1c is rapidly replaced by H1oo in a donor nucleus transplanted into an oocyte. The greater mobility of H1oo as compared to H1c may contribute to this rapid replacement and increased ... [full text in table]

Curated tissue note. Oocyte-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZA3 | HPA portal (live; not the pinned values)

H2AC1 | Unsafe / Protein

ENSG00000164508

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Supported); adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: Medium (Supported); caudate / glial cells: High (Supported); caudate / neuronal cells: High ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Colon Adenocarcinoma (validation): 1/486 (0.2%); Lung Adenocarcinoma (TCGA): 1/497 (0.2%)

Cancer IHC. glioma: 12/12 (100.0%); lung cancer: 11/11 (100.0%)

Biology. Core component of nucleosome. Nucleosomes wrap and compact DNA into chromatin, limiting DNA accessibility to the cellular machineries which require DNA as a template. Histones thereby play a central role in transcription regulation, DNA repair, DNA replication and chromosomal stability. DNA accessibility is regulated via a complex set of post-translational modifications of histones, also called histone code, and nucleosome remodeling

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96QV6 | HPA portal (live; not the pinned values)

H2BC12L | Unsafe / Protein

ENSG00000234289

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / glial cells: High (Supported); caudate / neuronal cells: Medium (Supported); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 15/58 (25.9%); Pancreatic Adenocarcinoma (validation): 6/80 (7.5%); Ovary Serous Cystadenocarcinoma (validation): 6/81 (7.4%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Core component of nucleosome. Nucleosomes wrap and compact DNA into chromatin, limiting DNA accessibility to the cellular machineries which require DNA as a template. Histones thereby play a central role in transcription regulation, DNA repair, DNA replication and chromosomal stability. DNA accessibility is regulated via a complex set of post-translational modifications of histones, also called histone code, and nucleosome remodeling ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P57053 | HPA portal (live; not the pinned values)

H2BC3 | Unsafe / Protein

ENSG00000276410

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / glial cells: High (Supported); caudate / neuronal cells: High (Supported); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 477/486 (98.1%); Rectum Adenocarcinoma (validation): 193/207 (93.2%); Colon Adenocarcinoma (TCGA): 6/254 (2.4%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Core component of nucleosome. Nucleosomes wrap and compact DNA into chromatin, limiting DNA accessibility to the cellular machineries which require DNA as a template. Histones thereby play a central role in transcription regulation, DNA repair, DNA replication and chromosomal stability. DNA accessibility is regulated via a complex set of post-translational modifications of histones, also called histone code, and nucleosome remodeling

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P33778 | HPA portal (live; not the pinned values)

H3-4 | Unsafe / Protein

ENSG00000168148

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: High (Approved); cerebellum ... [full text in table]

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 0/283 (0.0%)

Cancer IHC. glioma: 12/12 (100.0%); lymphoma: 12/12 (100.0%)

Biology. Core component of nucleosome. Nucleosomes wrap and compact DNA into chromatin, limiting DNA accessibility to the cellular machineries which require DNA as a template. Histones thereby play a central role in transcription regulation, DNA repair, DNA replication and chromosomal stability. DNA accessibility is regulated via a complex set of post-translational modifications of histones, also called histone code, and nucleosome remodeling

Curated tissue note. Expressed in testicular cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q16695 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

H3C12 | Unsafe / Protein

ENSG00000197153

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: High (Approved); cerebellum ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 485/486 (99.8%); Rectum Adenocarcinoma (validation): 203/207 (98.1%); Glioblastoma Multiforme (validation): 53/58 (91.4%)

Cancer IHC. glioma: 12/12 (100.0%); lymphoma: 12/12 (100.0%)

Biology. Core component of nucleosome. Nucleosomes wrap and compact DNA into chromatin, limiting DNA accessibility to the cellular machineries which require DNA as a template. Histones thereby play a central role in transcription regulation, DNA repair, DNA replication and chromosomal stability. DNA accessibility is regulated via a complex set of post-translational modifications of histones, also called histone code, and nucleosome remodeling

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P68431 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

H4C6 | Unsafe / Protein

ENSG00000274618

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: Medium (Supported); caudate / glial cells: High (Supported); cerebellum / cells in granular layer: Low (Supported); cerebellum / cells in molecular layer: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 483/486 (99.4%); Rectum Adenocarcinoma (validation): 202/207 (97.6%); Ovary Serous Cystadenocarcinoma (TCGA): 18/349 (5.2%)

Cancer IHC. breast cancer: 12/12 (100.0%); endometrial cancer: 12/12 (100.0%)

Biology. Core component of nucleosome. Nucleosomes wrap and compact DNA into chromatin, limiting DNA accessibility to the cellular machineries which require DNA as a template. Histones thereby play a central role in transcription regulation, DNA repair, DNA replication and chromosomal stability. DNA accessibility is regulated via a complex set of post-translational modifications of histones, also called histone code, and nucleosome remodeling

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P62805 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

HARBI1 | Unsafe / Protein

ENSG00000180423

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. stomach 1 / glandular cells: Medium (Approved); stomach 2 / glandular cells: Medium (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 44/50 (88.0%); Prostate Adenocarcinoma (TCGA): 414/480 (86.2%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Transposase-derived protein that may have nuclease activity (Potential). Does not have transposase activity

Curated tissue note. Detected in brain, eye, nerve tissue, kidney and lung

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96MB7 | HPA portal (live; not the pinned values)

HAUS6 | Unsafe / Protein

ENSG00000147874

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. colorectal cancer: 9/11 (81.8%); skin cancer: 9/12 (75.0%)

Biology. Contributes to mitotic spindle assembly, maintenance of centrosome integrity and completion of cytokinesis as part of the HAUS augmin-like complex. Promotes the nucleation of microtubules from the spindle through recruitment of NEDD1 and gamma-tubulin

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z4H7 | HPA portal (live; not the pinned values)

HBG2 | Unsafe / Protein

ENSG00000196565

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Approved); cerebellum / Purkinje cells: Low (Approved); liver / hepatocytes: Medium (Approved); lung / alveolar cells: Low (Approved); lung / macrophages: Low (Approved)

Cancer RNA. Glioblastoma Multiforme (TCGA): 48/141 (34.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 57/521 (10.9%); Breast Invasive Carcinoma (validation): 3/50 (6.0%)

Cancer IHC. liver cancer: 4/11 (36.4%); breast cancer: 0/12 (0.0%)

Biology. Gamma chains make up the fetal hemoglobin F, in combination with alpha chains

Curated tissue note. Red blood cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P69892 | HPA portal (live; not the pinned values)

HBZ | Unsafe / Protein

ENSG00000130656

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Approved); spleen / cells in red pulp: Medium (Approved)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Kidney Renal Clear Cell Carcinoma (TCGA): 2/521 (0.4%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. The zeta chain is an alpha-type chain of mammalian embryonic hemoglobin

Curated tissue note. Detected in fetal erythrocytes (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P02008 | HPA portal (live; not the pinned values)

HEATR4 | Unsafe / Protein

ENSG00000187105

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / peripheral nerve/ganglion: Low (Enhanced); kidney / proximal tubules (microvilli): High (Enhanced); parathyroid gland / glandular cells: Low (Enhanced); smooth muscle / smooth muscle cells: Low (Enhanced); stomach 1 / glandular cells: Low (Enhanced)

Cancer RNA. Colon Adenocarcinoma (validation): 77/486 (15.8%); Rectum Adenocarcinoma (validation): 31/207 (15.0%); Kidney Chromophobe (TCGA): 5/64 (7.8%)

Cancer IHC. stomach cancer: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86WZ0 | HPA portal (live; not the pinned values)

HEATR9 | Unsafe / High RNA

ENSG00000270379

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 4.8 nTPM; liver/b-cells; hpa_single_cell_tissue; vital; 55.93 TPM; blood/NK; dice_nk; vital

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A2RTY3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

HELB | Unsafe / High RNA

ENSG00000127311

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 56.3 nTPM; adipose tissue/nk-cells; hpa_single_cell_tissue; vital; 87.44 TPM; blood/TH2; dice_th2; vital

Cancer RNA. Glioblastoma Multiforme (validation): 50/58 (86.2%); Pancreatic Adenocarcinoma (validation): 59/80 (73.8%); Breast Invasive Carcinoma (validation): 22/50 (44.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. 5'-3' DNA helicase involved in DNA damage response by acting as an inhibitor of DNA end resection (PubMed:25617833, PubMed:26774285). Recruitment to single-stranded DNA (ssDNA) following DNA damage leads to inhibit the nucleases catalyzing resection, such as EXO1, BLM and DNA2, possibly via the 5'-3' ssDNA translocase activity of HELB (PubMed:26774285). As cells approach S phase, DNA end resection is promoted by the nuclear export of ... [full text in table]

Curated tissue note. Highly expressed in testis and thymus and weakly in liver, spleen, kidney and brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NG08 | HPA portal (live; not the pinned values)

HELQ | Unsafe / Protein

ENSG00000163312

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 77/80 (96.2%); Breast Invasive Carcinoma (validation): 45/50 (90.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); melanoma: 12/12 (100.0%)

Biology. Single-stranded 3'-5' DNA helicase that plays a key role in homology-driven double-strand break (DSB) repair (PubMed:11751861, PubMed:19995904, PubMed:21398521, PubMed:24005041, PubMed:24005565, PubMed:34316696, PubMed:34937945). Involved in different DSB repair mechanisms that are guided by annealing of extensive stretches of complementary bases at break ends, such as microhomology-mediated end-joining (MMEJ), single-strand annealing ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TDG4 | HPA portal (live; not the pinned values)

HEMGN | Unsafe / Protein

ENSG00000136929

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: High (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%); Skin Cuteneous Melanoma (TCGA): 1/99 (1.0%); Kidney Renal Clear Cell Carcinoma (validation): 1/100 (1.0%)

Cancer IHC. cervical cancer: 1/12 (8.3%); colorectal cancer: 1/12 (8.3%)

Biology. Regulates the proliferation and differentiation of hematopoietic cells. Overexpression block the TPA-induced megakaryocytic differentiation in the K562 cell model. May also prevent cell apoptosis through the activation of the nuclear factor-kappa B (NF-kB)

Curated tissue note. Expressed in hematopoietic precursor cells, thyroid and spermatids (at protein level). Expressed in bone marrow, testis, thymus. Expressed in prostate cancer and ovarian cancer. Also expressed in thymus and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BXL5 | HPA portal (live; not the pinned values)

HENMT1 | Unsafe / Protein

ENSG00000162639

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / Purkinje cells: Low (Uncertain); cerebral cortex / neuropil: High (Uncertain); colon / endothelial cells: Low (Uncertain); colon / glandular cells: Medium (Uncertain); colon / peripheral ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 205/207 (99.0%); Testicular Germ Cell Tumor (TCGA): 128/133 (96.2%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 272/283 (96.1%)

Cancer IHC. lymphoma: 10/12 (83.3%); testis cancer: 10/12 (83.3%)

Biology. Methyltransferase that adds a 2'-O-methyl group at the 3'-end of piRNAs, a class of 24 to 30 nucleotide RNAs that are generated by a Dicer-independent mechanism and are primarily derived from transposons and other repeated sequence elements. This probably protects the 3'-end of piRNAs from uridylation activity and subsequent degradation. Stabilization of piRNAs is essential for gametogenesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T8I9 | HPA portal (live; not the pinned values)

HERC5 | Unsafe / Protein

ENSG00000138646

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Supported); colon / glandular cells: Low (Supported); hippocampus / neuronal cells: Low (Supported); kidney / cells in tubules: Low (Supported); liver / hepatocytes: Low (Supported); rectum / glandular cells: Low (Supported); small intestine / glandular cells: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Lung Squamous Cell Carcinoma (validation): 61/68 (89.7%)

Cancer IHC. testis cancer: 6/11 (54.5%); ovarian cancer: 1/12 (8.3%)

Biology. Major E3 ligase for ISG15 conjugation (PubMed:16407192, PubMed:16815975, PubMed:16884686, PubMed:20308324, PubMed:20542004, PubMed:26355087, PubMed:27534820, PubMed:27564865, PubMed:37279284, PubMed:38421872, PubMed:38652662, PubMed:40184173). Functions as part of the ISGylation machinery that recognizes target proteins in a broad and relatively non-specific manner (PubMed:16407192, PubMed:16815975, PubMed:16884686, PubMed:20308324, ... [full text in table]

Curated tissue note. Expressed in testis and to a lesser degree in brain, ovary and placenta. Found in most tissues at low levels

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UII4 | HPA portal (live; not the pinned values)

HINT3 | Unsafe / Protein

ENSG00000111911

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bronchus / ciliated cells (cell body): Medium (Approved); bronchus / ciliated cells (ciliary rootlets): Medium (Approved); bronchus / ciliated cells (tip of cilia): Medium (Approved); caudate / glial ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); prostate cancer: 11/11 (100.0%)

Biology. Exhibits adenosine 5'-monophosphoramidase activity, hydrolyzing purine nucleotide phosphoramidates with a single phosphate group such as adenosine 5'monophosphoramidate (AMP-NH2) to yield AMP and NH2 (PubMed:17870088). Hydrolyzes lysyl-AMP (AMP-N-epsilon-(N-alpha-acetyl lysine methyl ester)) generated by lysine tRNA ligase (PubMed:17870088). Hydrolyzes 3-indolepropionic acyl-adenylate and fluorogenic purine nucleoside tryptamine ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NQE9 | HPA portal (live; not the pinned values)

HIPK3 | Unsafe / Protein

ENSG00000110422

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. colorectal cancer: 9/12 (75.0%); liver cancer: 7/12 (58.3%)

Biology. Serine/threonine-protein kinase involved in transcription regulation, apoptosis and steroidogenic gene expression. Phosphorylates JUN and RUNX2. Seems to negatively regulate apoptosis by promoting FADD phosphorylation. Enhances androgen receptor-mediated transcription. May act as a transcriptional corepressor for NK homeodomain transcription factors. The phosphorylation of NR5A1 activates SF1 leading to increased steroidogenic gene ... [full text in table]

Curated tissue note. Overexpressed in multidrug resistant cells. Highly expressed in heart and skeletal muscle, and at lower levels in placenta, pancreas, brain, spleen, prostate, thymus, testis, small intestine, colon and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H422 | HPA portal (live; not the pinned values)

HIPK4 | Unsafe / RNA

ENSG00000160396

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.9 nTPM (limit 2); basal ganglia: 8.8 nTPM (limit 2); cerebral cortex: 4.9 nTPM (limit 2); hippocampal formation: 4.1 nTPM (limit 2); amygdala: 2.9 nTPM (baseline 2); basal ganglia: 8.8 nTPM (baseline 2); cerebral cortex: 4.9 nTPM (baseline 2); hippocampal formation: 4.1 nTPM (baseline 2)

Cancer RNA. Kidney Chromophobe (TCGA): 1/64 (1.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 1/282 (0.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Protein kinase that phosphorylates human TP53 at Ser-9, and thus induces TP53 repression of BIRC5 promoter (By similarity). May act as a corepressor of transcription factors (Potential)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NE63 | HPA portal (live; not the pinned values)

HJURP | Unsafe / Protein

ENSG00000123485

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); cerebellum / Purkinje cells: Low (Uncertain); cerebral cortex / endothelial cells: Low (Uncertain); cerebral cortex / neuronal cells: Medium ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 483/486 (99.4%); Testicular Germ Cell Tumor (TCGA): 131/133 (98.5%); Rectum Adenocarcinoma (validation): 203/207 (98.1%)

Cancer IHC. colorectal cancer: 7/12 (58.3%); stomach cancer: 7/12 (58.3%)

Biology. Centromeric protein that plays a central role in the incorporation and maintenance of histone H3-like variant CENPA at centromeres. Acts as a specific chaperone for CENPA and is required for the incorporation of newly synthesized CENPA molecules into nucleosomes at replicated centromeres. Prevents CENPA-H4 tetramerization and prevents premature DNA binding by the CENPA-H4 tetramer. Directly binds Holliday junctions

Curated tissue note. According to PubMed:17256767, highly expressed in the thymus with lower levels in the placenta, small intestine, liver, skeletal muscle, and colon. According to PubMed:17823411, highly expressed in testis, and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCD3 | HPA portal (live; not the pinned values)

HMGA2 | Unsafe / Protein

ENSG00000149948

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 192/207 (92.8%); Colon Adenocarcinoma (validation): 435/486 (89.5%); Thyroid Carcinoma (TCGA): 390/495 (78.8%)

Cancer IHC. stomach cancer: 9/12 (75.0%); testis cancer: 8/11 (72.7%)

Biology. Functions as a transcriptional regulator. Functions in cell cycle regulation through CCNA2. Plays an important role in chromosome condensation during the meiotic G2/M transition of spermatocytes. Plays a role in postnatal myogenesis, is involved in satellite cell activation (By similarity). Positively regulates IGF2 expression through PLAG1 and in a PLAG1-independent manner (PubMed:28796236)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P52926 | HPA portal (live; not the pinned values)

HMGB4 | Unsafe / High RNA

ENSG00000176256

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 63.9 nTPM; testis/macrophages; hpa_single_cell_tissue; vital; 0.1165 TPM; blood/TREG_NAIVE; dice_treg_naive; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/9 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WW32 | HPA portal (live; not the pinned values)

HMHB1 | Unsafe / High RNA

ENSG00000158497

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 31.9 nTPM; blood/plasmacytoid DC; hpa_immune_cell_samples; vital; 30.08 TPM; blood/plasmacytoid DC; hpa_immune_cell_samples; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 1/50 (2.0%); Glioblastoma Multiforme (TCGA): 1/141 (0.7%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Precursor of the histocompatibility antigen HB-1. More generally, minor histocompatibility antigens (mHags) refer to immunogenic peptide which, when complexed with MHC, can generate an immune response after recognition by specific T-cells. The peptides are derived from polymorphic intracellular proteins, which are cleaved by normal pathways of antigen processing. The binding of these peptides to MHC class I or class II molecules and ... [full text in table]

Curated tissue note. Expressed in acute lymphoblastic leukemia B-cells and Epstein-Barr virus-transformed B-cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O97980 | HPA portal (live; not the pinned values)

HMMR | Unsafe / Protein

ENSG00000072571

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); colon / glandular cells: Low (Enhanced); duodenum / glandular cells: Low (Enhanced); hippocampus / neuronal cells: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 482/486 (99.2%); Colon Adenocarcinoma (TCGA): 246/254 (96.9%); Glioblastoma Multiforme (validation): 56/58 (96.6%)

Cancer IHC. breast cancer: 3/12 (25.0%); colorectal cancer: 1/10 (10.0%)

Biology. Receptor for hyaluronic acid (HA) (By similarity). Involved in cell motility (By similarity). When hyaluronan binds to HMMR, the phosphorylation of a number of proteins, including PTK2/FAK1 occurs. May also be involved in cellular transformation and metastasis formation, and in regulating extracellular-regulated kinase (ERK) activity. May act as a regulator of adipogenisis (By similarity)

Curated tissue note. Expressed in testis (PubMed:22965910). Expressed in the breast (PubMed:8890751)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75330 | HPA portal (live; not the pinned values)

HROB | Unsafe / Protein

ENSG00000125319

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 114/133 (85.7%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 219/283 (77.4%); Lung Squamous Cell Carcinoma (TCGA): 297/489 (60.7%)

Cancer IHC. prostate cancer: 11/12 (91.7%); colorectal cancer: 9/11 (81.8%)

Biology. DNA-binding protein involved in homologous recombination that acts by recruiting the MCM8-MCM9 helicase complex to sites of DNA damage to promote DNA repair synthesis (PubMed:31467087). A C-terminal region including the OB-fold stimulates the helicase activity of MCM8-MCM9 probably by altering its conformation (PubMed:37535404)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N3J3 | HPA portal (live; not the pinned values)

HS6ST3 | Unsafe / RNA

ENSG00000185352

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.1 nTPM (limit 2); amygdala: 5.2 nTPM (limit 2); basal ganglia: 5.9 nTPM (limit 2); cerebral cortex: 16.7 nTPM (limit 2); hippocampal formation: 10.9 nTPM (limit 2); hypothalamus: 6.7 nTPM (limit 2); midbrain: 3 nTPM (limit 2); pancreas: 2.1 nTPM (limit 2); retina: 2.2 nTPM (limit ... [full text in table]

Cancer RNA. Kidney Chromophobe (TCGA): 55/64 (85.9%); Breast Invasive Carcinoma (validation): 24/50 (48.0%); Glioblastoma Multiforme (validation): 16/58 (27.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. 6-O-sulfation enzyme which catalyzes the transfer of sulfate from 3'-phosphoadenosine 5'-phosphosulfate (PAPS) to position 6 of the N-sulfoglucosamine residue (GlcNS) of heparan sulfate

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZP7 | HPA portal (live; not the pinned values)

HSD17B1 | Unsafe / RNA

ENSG00000108786

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3 nTPM (limit 2); basal ganglia: 3.1 nTPM (limit 2); choroid plexus: 3 nTPM (limit 2); hippocampal formation: 3.4 nTPM (limit 2); hypothalamus: 2.4 nTPM (limit 2); midbrain: 3.4 nTPM (limit 2); pituitary gland: 2.3 nTPM (limit 2); spinal cord: 3 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 48/50 (96.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 457/492 (92.9%); Ovary Serous Cystadenocarcinoma (TCGA): 310/349 (88.8%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Favors the reduction of estrogens and androgens. Converts estrone (E1) to a more potent estrogen, 17beta-estradiol (E2) (PubMed:8994190). Also has 20-alpha-HSD activity. Uses preferentially NADH

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P14061 | HPA portal (live; not the pinned values)

HSD17B3 | Unsafe / RNA

ENSG00000130948

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.8 nTPM (limit 2); basal ganglia: 3.2 nTPM (limit 2); hippocampal formation: 4.1 nTPM (limit 2); hypothalamus: 2.2 nTPM (limit 2); liver: 14.8 nTPM (limit 12); midbrain: 4.2 nTPM (limit 2); spinal cord: 5 nTPM (limit 2)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 172/231 (74.5%); Glioblastoma Multiforme (validation): 26/58 (44.8%); Breast Invasive Carcinoma (validation): 8/50 (16.0%)

Cancer IHC. liver cancer: 5/12 (41.7%); lung cancer: 4/12 (33.3%)

Biology. Catalyzes the conversion of 17-oxosteroids to 17beta-hydroxysteroids (PubMed:16216911, PubMed:23856005, PubMed:26545797, PubMed:27927697, PubMed:8075637). Favors the reduction of androstenedione to testosterone (PubMed:16216911, PubMed:23856005, PubMed:26545797, PubMed:27927697). Testosterone is the key androgen driving male development and function (PubMed:8075637). Uses NADPH while the two other EDH17B enzymes use NADH ... [full text in table]

Curated tissue note. Testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P37058 | HPA portal (live; not the pinned values)

HSD3B1 | Unsafe / Protein

ENSG00000203857

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / cells in zona fasciculata: Low (Supported); adrenal gland / cells in zona glomerulosa: Low (Supported); adrenal gland / glandular cells: Low (Supported)

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 13/169 (7.7%); Testicular Germ Cell Tumor (TCGA): 6/133 (4.5%); Liver Hepatocellular Carcinoma (TCGA): 11/362 (3.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. A bifunctional enzyme responsible for the oxidation and isomerization of 3beta-hydroxy-Delta(5)-steroid precursors to 3-oxo-Delta(4)-steroids, an essential step in steroid hormone biosynthesis. Specifically catalyzes the conversion of pregnenolone to progesterone, 17alpha-hydroxypregnenolone to 17alpha-hydroxyprogesterone, dehydroepiandrosterone (DHEA) to 4-androstenedione, and androstenediol to testosterone. Additionally, catalyzes ... [full text in table]

Curated tissue note. Placenta and skin (PubMed:1401999). Predominantly expressed in mammary gland tissue

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P14060 | HPA portal (live; not the pinned values)

HSF2 | Unsafe / RNA

ENSG00000025156

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 14.7 nTPM (limit 4); amygdala: 8.5 nTPM (limit 2); basal ganglia: 8.7 nTPM (limit 2); bone marrow: 29.2 nTPM (limit 4); cerebellum: 17.9 nTPM (limit 4); cerebral cortex: 11.5 nTPM (limit 4); choroid plexus: 16.1 nTPM (limit 2); colon: 11.2 nTPM (limit 8); duodenum: 6.7 nTPM (limit ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. testis cancer: 5/11 (45.5%); breast cancer: 0/12 (0.0%)

Biology. DNA-binding protein that specifically binds heat shock promoter elements (HSE) and activates transcription. In higher eukaryotes, HSF is unable to bind to the HSE unless the cells are heat shocked

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q03933 | HPA portal (live; not the pinned values)

HSF2BP | Unsafe / Protein

ENSG00000160207

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); hippocampus / neuronal cells: Low (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 37/58 (63.8%); Breast Invasive Carcinoma (validation): 31/50 (62.0%); Bladder Urothelial Carcinoma (TCGA): 14/169 (8.3%)

Cancer IHC. breast cancer: 1/12 (8.3%); cervical cancer: 0/11 (0.0%)

Biology. Meiotic recombination factor component of recombination bridges involved in meiotic double-strand break repair. Modulates the localization of recombinases DMC1:RAD51 to meiotic double-strand break (DSB) sites through the interaction with BRCA2 and its recruitment during meiotic recombination (By similarity) (PubMed:31242413). Indispensable for the DSB repair, homologous synapsis, and crossover formation that are needed for progression ... [full text in table]

Curated tissue note. Testis specific. Overexpressed in some tumors (PubMed:31242413)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75031 | HPA portal (live; not the pinned values)

HSFX4 | Unsafe / RNA

ENSG00000283463

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.7 nTPM (limit 2); adrenal gland: 2.7 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 9/176 (5.1%); Kidney Chromophobe (TCGA): 1/64 (1.6%); Prostate Adenocarcinoma (TCGA): 5/480 (1.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0A1B0GTS1 | HPA portal (live; not the pinned values)

HSPA1L | Unsafe / Protein

ENSG00000204390

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Enhanced); nasopharynx / ciliated cells (cell body): Medium (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 45/50 (90.0%); Glioblastoma Multiforme (validation): 50/58 (86.2%); Kidney Chromophobe (TCGA): 1/64 (1.6%)

Cancer IHC. ovarian cancer: 1/11 (9.1%); breast cancer: 0/10 (0.0%)

Biology. Molecular chaperone implicated in a wide variety of cellular processes, including protection of the proteome from stress, folding and transport of newly synthesized polypeptides, activation of proteolysis of misfolded proteins and the formation and dissociation of protein complexes. Plays a pivotal role in the protein quality control system, ensuring the correct folding of proteins, the re-folding of misfolded proteins and controlling ... [full text in table]

Curated tissue note. Expressed in spermatids

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P34931 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

HSPA4L | Unsafe / Protein

ENSG00000164070

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebellum / Purkinje cells - cytoplasm/membrane: Medium (Enhanced); cerebellum / Purkinje cells - dendrites: Medium (Enhanced); lung / macrophages: Low (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 95/100 (95.0%); Kidney Chromophobe (TCGA): 59/64 (92.2%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/9 (0.0%)

Biology. Possesses chaperone activity in vitro where it inhibits aggregation of citrate synthase

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95757 | HPA portal (live; not the pinned values)

HTN1 | Unsafe / RNA

ENSG00000126550

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. heart muscle: 4.3 nTPM (limit 4); pancreas: 18 nTPM (limit 4); salivary gland: 47123.7 nTPM (limit 8)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 20/80 (25.0%); Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%); Ovary Serous Cystadenocarcinoma (validation): 1/81 (1.2%)

Cancer IHC. skin cancer: 9/12 (75.0%); urothelial cancer: 5/12 (41.7%)

Biology. Histatins (Hsts) are cationic and histidine-rich secreted peptides mainly synthesized by saliva glands of humans and higher primates (PubMed:3286634, PubMed:3944083). Hsts are considered to be major precursors of the protective proteinaceous structure on tooth surfaces (enamel pellicle). Hsts can be divided into two major groups according to their biological functions: antimicrobial Hsts (e.g. Hst 5/HTN3) and cell-activating Hsts (e.g. ... [full text in table]

Curated tissue note. Submandibular and parotid glands

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P15515 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

HTN3 | Unsafe / Protein

ENSG00000205649

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. salivary gland / glandular cells: High (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (validation): 50/207 (24.2%); Colon Adenocarcinoma (validation): 70/486 (14.4%); Pancreatic Adenocarcinoma (validation): 5/80 (6.2%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Histatins are cationic and histidine-rich peptides mainly found in the saliva of higher primates (PubMed:3286634). They are considered to be major precursors of the protective proteinaceous structure on tooth surfaces (enamel pellicle). Hsts can be divided into two major groups according to their biological functions: antimicrobial Hsts (e.g. Hst 5/HTN3) and cell-activating Hsts (e.g. Hst 1/HTN1, Hst 2/HTN1 and Hst 3/HTN3) ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P15516 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

HTR7 | Unsafe / Protein

ENSG00000148680

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); cerebral cortex / glial cells: Medium (Approved); cerebral cortex / neuronal cells: Medium (Approved); cerebral cortex / neuronal projections: Medium (Approved); colon / glandular cells: Low ... [full text in table]

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 182/492 (37.0%); Lung Squamous Cell Carcinoma (validation): 13/68 (19.1%); Bladder Urothelial Carcinoma (TCGA): 31/169 (18.3%)

Cancer IHC. liver cancer: 4/11 (36.4%); endometrial cancer: 4/12 (33.3%)

Biology. G protein-coupled receptor for 5-hydroxytryptamine (serotonin), a biogenic hormone that functions as a neurotransmitter, a hormone and a mitogen (PubMed:35714614, PubMed:8226867). Ligand binding causes a conformation change that triggers signaling via guanine nucleotide-binding proteins (G proteins) and modulates the activity of downstream effectors (PubMed:35714614, PubMed:8226867). HTR7 is coupled to G(s) G alpha proteins and ... [full text in table]

Curated tissue note. Predominant isoform in spleen, caudate and hippocampus Expressed at lower levels Minor isoform in terms of expression

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P34969 | HPA portal (live; not the pinned values)

HUNK | Unsafe / Protein

ENSG00000142149

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 178/207 (86.0%); Colon Adenocarcinoma (validation): 351/486 (72.2%); Testicular Germ Cell Tumor (TCGA): 86/133 (64.7%)

Cancer IHC. testis cancer: 8/9 (88.9%); endometrial cancer: 6/12 (50.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P57058 | HPA portal (live; not the pinned values)

HYCC1 | Unsafe / Protein

ENSG00000122591

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / basal cells: Medium (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / goblet cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Adenocarcinoma (validation): 104/105 (99.0%); Lung Squamous Cell Carcinoma (validation): 65/68 (95.6%)

Cancer IHC. colorectal cancer: 5/11 (45.5%); testis cancer: 5/12 (41.7%)

Biology. Component of a complex required to localize phosphatidylinositol 4-kinase (PI4K) to the plasma membrane (PubMed:26571211). The complex acts as a regulator of phosphatidylinositol 4-phosphate (PtdIns(4)P) synthesis (PubMed:26571211). HYCC1 plays a key role in oligodendrocytes formation, a cell type with expanded plasma membrane that requires generation of PtdIns(4)P (PubMed:26571211). Its role in oligodendrocytes formation probably ... [full text in table]

Curated tissue note. Widely expressed. Highest levels in heart, brain, placenta, spleen and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BYI3 | HPA portal (live; not the pinned values)

HYDIN | Unsafe / Protein

ENSG00000157423

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. nasopharynx / ciliated cells (tip of cilia): Low (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 54/58 (93.1%); Breast Invasive Carcinoma (validation): 37/50 (74.0%); Pancreatic Adenocarcinoma (validation): 28/80 (35.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Required for ciliary motility (By similarity). Required for localization of SPEF2 to the ciliary axoneme in ciliated respiratory epithelial cells (PubMed:31545650)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4G0P3 | HPA portal (live; not the pinned values)

IBA57 | Unsafe / Protein

ENSG00000181873

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: Low (Approved); colon / peripheral nerve/ganglion: Low (Approved); kidney / cells in tubules: Medium (Approved); lung / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 45/58 (77.6%); Colon Adenocarcinoma (validation): 187/486 (38.5%); Pancreatic Adenocarcinoma (validation): 30/80 (37.5%)

Cancer IHC. prostate cancer: 11/11 (100.0%); melanoma: 7/10 (70.0%)

Biology. Mitochondrial protein involved in the maturation of mitochondrial [4Fe-4S]-proteins in the late stage of the iron-sulfur cluster assembly pathway (PubMed:22323289, PubMed:23462291). Operates in cooperation with ISCA2 in the maturation of [4Fe-4S] proteins (PubMed:30269484) Involved in the maturation of mitochondrial 2Fe-2S proteins in the late stage of the iron-sulfur cluster assembly pathway

Curated tissue note. Expressed in skin fibroblasts and skeletal muscle (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T440 | HPA portal (live; not the pinned values)

IER3IP1 | Unsafe / Protein

ENSG00000134049

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: Low (Supported); bronchus / basal cells: Medium (Supported); bronchus / ciliated cells (cell body): Medium (Supported); bronchus / goblet cells: Medium (Supported); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); endometrial cancer: 10/11 (90.9%)

Biology. Regulator of endoplasmic reticulum secretion that acts as a key determinant of brain size (PubMed:33122427). Required for secretion of extracellular matrix proteins (PubMed:33122427). Required for correct brain development by depositing sufficient extracellular matrix proteins for tissue integrity and the proliferation of neural progenitors (PubMed:33122427). Acts as a regulator of the unfolded protein response (UPR) (By similarity)

Curated tissue note. Highest levels in heart, skeletal muscle, and kidney

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5U9 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

IFT56 | Unsafe / Protein

ENSG00000105948

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Low (Enhanced); bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); cerebral cortex / neuropil: Low (Enhanced); nasopharynx ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Kidney Renal Clear Cell Carcinoma (validation): 82/100 (82.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Component of the intraflagellar transport (IFT) complex B required for transport of proteins in the motile cilium. Required for transport of specific ciliary cargo proteins related to motility, while it is neither required for IFT complex B assembly or motion nor for cilium assembly. Required for efficient coupling between the accumulation of GLI2 and GLI3 at the ciliary tips and their dissociation from the negative regulator SUFU. ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0AVF1 | HPA portal (live; not the pinned values)

IFT70A | Unsafe / Protein

ENSG00000197557

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Supported); nasopharynx / respiratory epithelial cells: Medium (Supported)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Pancreatic Adenocarcinoma (validation): 63/80 (78.8%)

Cancer IHC. liver cancer: 2/11 (18.2%); lung cancer: 2/12 (16.7%)

Biology. Required for polyglutamylation of axonemal tubulin. Plays a role in anterograde intraflagellar transport (IFT), the process by which cilia precursors are transported from the base of the cilium to the site of their incorporation at the tip

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86WT1 | HPA portal (live; not the pinned values)

IFT70B | Unsafe / Protein

ENSG00000196659

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: High (Supported); nasopharynx / respiratory epithelial cells: Medium (Supported)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 47/50 (94.0%); Ovary Serous Cystadenocarcinoma (TCGA): 270/349 (77.4%)

Cancer IHC. liver cancer: 2/11 (18.2%); lung cancer: 2/12 (16.7%)

Biology. Required for polyglutamylation of axonemal tubulin. Plays a role in anterograde intraflagellar transport (IFT), the process by which cilia precursors are transported from the base of the cilium to the site of their incorporation at the tip

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N4P2 | HPA portal (live; not the pinned values)

IGF2BP1 | Unsafe / Protein

ENSG00000159217

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced); kidney / collecting ducts: Medium (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 129/133 (97.0%); Liver Hepatocellular Carcinoma (validation): 59/231 (25.5%); Lung Squamous Cell Carcinoma (TCGA): 101/489 (20.7%)

Cancer IHC. testis cancer: 12/12 (100.0%); colorectal cancer: 4/9 (44.4%)

Biology. RNA-binding factor that recruits target transcripts to cytoplasmic protein-RNA complexes (mRNPs). This transcript 'caging' into mRNPs allows mRNA transport and transient storage. It also modulates the rate and location at which target transcripts encounter the translational apparatus and shields them from endonuclease attacks or microRNA-mediated degradation. Preferentially binds to N6-methyladenosine (m6A)-containing mRNAs and ... [full text in table]

Curated tissue note. Mainly expressed in the embryo, including in fetal liver, fetal lung, fetal kidney, fetal thymus (at protein level). Also expressed follicles of ovary, as well as in gonocytes of testis, spermatogonia, semen, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NZI8 | HPA portal (live; not the pinned values)

IGF2BP3 | Unsafe / Protein

ENSG00000136231

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. 209.2 nTPM; pbmc/platelets; hpa_single_cell_tissue; vital; 2.6 TPM; blood/Progenitor cell; hpa_immune_monaco; vital

Cancer RNA. Glioblastoma Multiforme (validation): 50/58 (86.2%); Pancreatic Adenocarcinoma (validation): 67/80 (83.8%); Lung Squamous Cell Carcinoma (validation): 53/68 (77.9%)

Cancer IHC. lung cancer: 7/12 (58.3%); pancreatic cancer: 6/12 (50.0%)

Biology. RNA-binding factor that may recruit target transcripts to cytoplasmic protein-RNA complexes (mRNPs). This transcript 'caging' into mRNPs allows mRNA transport and transient storage. It also modulates the rate and location at which target transcripts encounter the translational apparatus and shields them from endonuclease attacks or microRNA-mediated degradation. Preferentially binds to N6-methyladenosine (m6A)-containing mRNAs and ... [full text in table]

Curated tissue note. Expressed in fetal liver, fetal lung, fetal kidney, fetal thymus, fetal placenta, fetal follicles of ovary and gonocytes of testis, growing oocytes, spermatogonia and semen (at protein level). Expressed in ... [full text in table]

Therapeutic evidence. Clinical publication.

UniProt O00425 | Trial/source record | HPA portal (live; not the pinned values)

IGFLR1 | Unsafe / Protein

ENSG00000126246

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: High (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 490/497 (98.6%); Glioblastoma Multiforme (TCGA): 139/141 (98.6%); Liver Hepatocellular Carcinoma (validation): 219/231 (94.8%)

Cancer IHC. liver cancer: 2/8 (25.0%); glioma: 1/11 (9.1%)

Biology. Probable cell membrane receptor for the IGF-like family proteins. Binds IGFL1 and IGFL3 with a higher affinity. May also bind IGFL2

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H665 | HPA portal (live; not the pinned values)

IGLL1 | Unsafe / Protein

ENSG00000128322

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 8/133 (6.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 15/283 (5.3%); Kidney Renal Papillary Cell Carcinoma (TCGA): 9/282 (3.2%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Critical for B-cell development

Curated tissue note. Expressed only in pre-B-cells and a special B-cell line (which is surface Ig negative)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P15814 | HPA portal (live; not the pinned values)

IKZF3 | Unsafe / Protein

ENSG00000161405

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); colon / mucosal lymphoid cells: High (Enhanced); lung / alveolar cells: Low (Enhanced); lymph node / non-germinal center cells: High (Enhanced); rectum / mucosal lymphoid cells: High (Enhanced); spleen / cells in white pulp: High (Enhanced)

Cancer RNA. Colon Adenocarcinoma (validation): 447/486 (92.0%); Rectum Adenocarcinoma (validation): 186/207 (89.9%); Lung Adenocarcinoma (validation): 82/105 (78.1%)

Cancer IHC. lymphoma: 11/12 (91.7%); glioma: 1/11 (9.1%)

Biology. Transcription factor that plays an important role in the regulation of lymphocyte differentiation. Plays an essential role in regulation of B-cell differentiation, proliferation and maturation to an effector state. Involved in regulating BCL2 expression and controlling apoptosis in T-cells in an IL2-dependent manner

Curated tissue note. Expressed most strongly in peripheral blood leukocytes, the spleen, and the thymus

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UKT9 | HPA portal (live; not the pinned values)

IL12RB2 | Unsafe / RNA

ENSG00000081985

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pancreas: 2.5 nTPM (limit 2); skeletal muscle: 3.4 nTPM (limit 2); pancreas: 2.5 nTPM (baseline 2); skeletal muscle: 3.4 nTPM (baseline 2)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 71/99 (71.7%); Head and Neck Squamous Cell Carcinoma (TCGA): 216/492 (43.9%); Kidney Renal Papillary Cell Carcinoma (TCGA): 107/282 (37.9%)

Cancer IHC. stomach cancer: 10/12 (83.3%); liver cancer: 7/12 (58.3%)

Biology. Receptor for interleukin-12. This subunit is the signaling component coupling to the JAK2/STAT4 pathway. Promotes the proliferation of T-cells as well as NK cells. Induces the promotion of T-cells towards the Th1 phenotype by strongly enhancing IFN-gamma production

Curated tissue note. Isoform 2 is expressed at similar levels in both naive and activated T-cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q99665 | HPA portal (live; not the pinned values)

IL13 | Unsafe / Normal

ENSG00000169194

Decision. Published protein production in non-permitted normal cells.

Normal RNA / IHC. 336.5 nTPM; fallopian tube/granulocytes; hpa_single_cell_tissue; vital; 18.28 TPM; blood/CD8_STIM; dice_cd8_stim; vital

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Liver Hepatocellular Carcinoma (TCGA): 1/362 (0.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Cytokine that plays important roles in allergic inflammation and immune response to parasite infection (PubMed:8096327, PubMed:8097324). Synergizes with IL2 in regulating interferon-gamma synthesis (PubMed:8096327). Stimulates B-cell proliferation, and activation of eosinophils, basophils, and mast cells (PubMed:7903680, PubMed:8759755). Plays an important role in controlling IL33 activity by modulating the production of transmembrane ... [full text in table]

Therapeutic evidence. Normal function.

UniProt P35225 | Reviewed source 1 | HPA portal (live; not the pinned values)

IL13RA2 | Unsafe / RNA

ENSG00000123496

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.6 nTPM (limit 2); amygdala: 2.8 nTPM (limit 2); basal ganglia: 5.6 nTPM (limit 2); cerebral cortex: 2.3 nTPM (limit 2); choroid plexus: 8.7 nTPM (limit 2); hippocampal formation: 4 nTPM (limit 2); hypothalamus: 11.7 nTPM (limit 2); kidney: 3.5 nTPM (limit 2); liver: 9.7 nTPM (limit ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 47/58 (81.0%); Glioblastoma Multiforme (TCGA): 111/141 (78.7%); Skin Cuteneous Melanoma (TCGA): 30/99 (30.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Cell surface receptor that plays a role in the regulation of IL-13-mediated responses (PubMed:11861389, PubMed:17030238). Functions as a decoy receptor that inhibits IL-13- and IL-4-mediated signal transduction via the JAK-STAT pathway and thereby modulates immune responses and inflammation (PubMed:11861389, PubMed:17030238). Serves as a functional signaling receptor for IL-13 in an alternative pathway involving AP-1 ultimately leading ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14627 | HPA portal (live; not the pinned values)

IL17F | Unsafe / High RNA

ENSG00000112116

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 107.2 nTPM; tongue/t-cells; hpa_single_cell_tissue; vital; 4.083 TPM; blood/CD4_STIM; dice_cd4_stim; vital

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 2/169 (1.2%); Rectum Adenocarcinoma (validation): 2/207 (1.0%); Stomach Adenocarcinoma (TCGA): 2/346 (0.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Effector cytokine of innate and adaptive immune system involved in antimicrobial host defense and maintenance of tissue integrity (PubMed:21350122). IL17A-IL17F signals via IL17RA-IL17RC heterodimeric receptor complex, triggering homotypic interaction of IL17RA and IL17RC chains with TRAF3IP2 adapter through SEFIR domains. This leads to downstream TRAF6-mediated activation of NF-kappa-B and MAPkinase pathways ultimately resulting in ... [full text in table]

Curated tissue note. Expressed in T-helper 1 and T-helper 2 cells, basophils and mast cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96PD4 | HPA portal (live; not the pinned values)

IL1A | Unsafe / RNA

ENSG00000115008

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. esophagus: 12.9 nTPM (limit 6); lung: 2.3 nTPM (limit 2); urinary bladder: 25 nTPM (limit 6); esophagus: 12.9 nTPM (baseline 6); lung: 2.3 nTPM (baseline 2); urinary bladder: 25 nTPM (baseline 6)

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 371/492 (75.4%); Lung Squamous Cell Carcinoma (validation): 43/68 (63.2%); Glioblastoma Multiforme (validation): 31/58 (53.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Cytokine constitutively present intracellularly in nearly all resting non-hematopoietic cells that plays an important role in inflammation and bridges the innate and adaptive immune systems (PubMed:26439902). After binding to its receptor IL1R1 together with its accessory protein IL1RAP, forms the high affinity interleukin-1 receptor complex (PubMed:17507369, PubMed:2950091). Signaling involves the recruitment of adapter molecules such ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P01583 | HPA portal (live; not the pinned values)

IL21 | Unsafe / High RNA

ENSG00000138684

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 15.8 nTPM; stomach/t-cells; hpa_single_cell_tissue; vital; 248 TPM; blood/CD4_STIM; dice_cd4_stim; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 1/50 (2.0%); Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Cytokine with immunoregulatory activity. May promote the transition between innate and adaptive immunity. Induces the production of IgG(1) and IgG(3) in B-cells (By similarity). Implicated in the generation and maintenance of T follicular helper (Tfh) cells and the formation of germinal-centers. Together with IL6, control the early generation of Tfh cells and are critical for an effective antibody response to acute viral infection (By ... [full text in table]

Curated tissue note. Expressed in activated CD4-positive T-cells but not in CD8-positive T-cells, B-cells, or monocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HBE4 | HPA portal (live; not the pinned values)

IL3 | Unsafe / High RNA

ENSG00000164399

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 12.4 nTPM; breast/t-cells; hpa_single_cell_tissue; vital; 78.67 TPM; blood/CD4_STIM; dice_cd4_stim; vital

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Cytokine secreted predominantly by activated T-lymphocytes as well as mast cells and osteoblastic cells that controls the production and differentiation of hematopoietic progenitor cells into lineage-restricted cells (PubMed:2556442). Also stimulates mature basophils, eosinophils, and monocytes to become functionally activated (PubMed:10779277, PubMed:32889153). In addition, plays an important role in neural cell proliferation and ... [full text in table]

Curated tissue note. Activated T-cells, mast cells, natural killer cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P08700 | HPA portal (live; not the pinned values)

IL4I1 | Unsafe / Protein

ENSG00000104951

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / basal cells: Medium (Enhanced); bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); bronchus / goblet cells: Medium (Enhanced); cerebellum / Purkinje cells: Low ... [full text in table]

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 328/349 (94.0%); Ovary Serous Cystadenocarcinoma (validation): 71/81 (87.7%); Kidney Renal Clear Cell Carcinoma (TCGA): 452/521 (86.8%)

Cancer IHC. lymphoma: 9/12 (75.0%); glioma: 6/12 (50.0%)

Biology. Secreted L-amino-acid oxidase that acts as a key immunoregulator (PubMed:17356132, PubMed:32818467, PubMed:32866000). Has preference for L-aromatic amino acids: converts phenylalanine (Phe), tyrosine (Tyr) and tryptophan (Trp) to phenylpyruvic acid (PP), hydroxyphenylpyruvic acid (HPP), and indole-3-pyruvic acid (I3P), respectively (PubMed:17356132, PubMed:32818467, PubMed:32866000). Also has weak L-arginine oxidase activity ... [full text in table]

Curated tissue note. Primarily found in immune tissues, with the highest expression in lymph nodes and spleen (PubMed:12031486, PubMed:12446450). Present in germinal center macrophages and inflammatory myeloid cells and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96RQ9 | HPA portal (live; not the pinned values)

IL5 | Unsafe / Normal

ENSG00000113525

Decision. Published protein production in non-permitted normal cells.

Normal RNA / IHC. 22.4 nTPM; skin/t-cells; hpa_single_cell_tissue; vital; 10.17 TPM; blood/CD8_STIM; dice_cd8_stim; vital

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 5/283 (1.8%); Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Homodimeric cytokine expressed predominantly by T-lymphocytes and NK cells that plays an important role in the survival, differentiation, and chemotaxis of eosinophils (PubMed:2653458, PubMed:9010276). Also acts on activated and resting B-cells to induce immunoglobulin production, growth, and differentiation (By similarity). Mechanistically, exerts its biological effects through a receptor composed of IL5RA subunit and the cytokine ... [full text in table]

Curated tissue note. Present in peripheral blood mononuclear cells

Therapeutic evidence. Normal function.

UniProt P05113 | Reviewed source 1 | HPA portal (live; not the pinned values)

IL7 | Unsafe / Protein

ENSG00000104432

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / basal cells: High (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / goblet cells: High (Approved); nasopharynx / basal cells: High (Approved); nasopharynx / ciliated cells (cell body): High (Approved); nasopharynx / ciliated cells (tip of cilia): Low ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 63/80 (78.8%); Colon Adenocarcinoma (validation): 329/486 (67.7%); Kidney Renal Clear Cell Carcinoma (validation): 67/100 (67.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); breast cancer: 11/11 (100.0%)

Biology. Hematopoietic cytokine that plays an essential role in the development, expansion, and survival of naive and memory T-cells and B-cells thereby regulating the number of mature lymphocytes and maintaining lymphoid homeostasis (PubMed:25870237, PubMed:7527823). Mechanistically, exerts its biological effects through a receptor composed of IL7RA subunit and the cytokine receptor common subunit gamma/CSF2RG (PubMed:8128231). Binding to the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P13232 | HPA portal (live; not the pinned values)

IMMP1L | Unsafe / Protein

ENSG00000148950

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%); Pancreatic Adenocarcinoma (TCGA): 176/176 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. liver cancer: 7/8 (87.5%); melanoma: 7/10 (70.0%)

Biology. Catalyzes the removal of transit peptides required for the targeting of proteins from the mitochondrial matrix, across the inner membrane, into the inter-membrane space. Known to process the nuclear encoded protein DIABLO

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96LU5 | HPA portal (live; not the pinned values)

IMPACT | Unsafe / Protein

ENSG00000154059

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / endothelial cells: Low ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 105/105 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. breast cancer: 9/11 (81.8%); testis cancer: 9/11 (81.8%)

Biology. Translational regulator that ensures constant high levels of translation upon a variety of stress conditions, such as amino acid starvation, UV-C irradiation, proteasome inhibitor treatment and glucose deprivation. Plays a role as a negative regulator of the EIF2AK4/GCN2 kinase activity; impairs GCN1-mediated EIF2AK4/GCN2 activation, and hence EIF2AK4/GCN2-mediated eIF-2-alpha phosphorylation and subsequent down-regulation of protein ... [full text in table]

Curated tissue note. Widely expressed. Expressed at high level in brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P2X3 | HPA portal (live; not the pinned values)

INCA1 | Unsafe / RNA

ENSG00000196388

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.6 nTPM (limit 2); amygdala: 2.9 nTPM (limit 2); basal ganglia: 3 nTPM (limit 2); cerebellum: 2.6 nTPM (limit 2); cerebral cortex: 6.9 nTPM (limit 2); choroid plexus: 3.3 nTPM (limit 2); duodenum: 10.4 nTPM (limit 2); heart muscle: 6.2 nTPM (limit 2); hippocampal formation: 4 nTPM ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 92/105 (87.6%); Rectum Adenocarcinoma (validation): 181/207 (87.4%); Kidney Renal Clear Cell Carcinoma (validation): 85/100 (85.0%)

Cancer IHC. colorectal cancer: 11/12 (91.7%); testis cancer: 9/10 (90.0%)

Biology. Binds to CDK2-bound cyclins and inhibits the kinase activity of CDK2; binding to cyclins is critical for its function as CDK inhibitor (PubMed:21540187). Inhibits cell growth and cell proliferation and may play a role in cell cycle control (By similarity). Required for ING5-mediated regulation of S-phase progression, enhancement of Fas-induced apoptosis and inhibition of cell growth (By similarity)

Curated tissue note. Detected in testis, and at lower levels in ovary. Detected at very low levels in testis tumors (PubMed:15159402). Down-regulated in bone marrow cells in acute myeloid and lymphoid leukemia patients as compared ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q0VD86 | HPA portal (live; not the pinned values)

INHA | Unsafe / Protein

ENSG00000123999

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 33/50 (66.0%); Testicular Germ Cell Tumor (TCGA): 36/133 (27.1%); Lung Adenocarcinoma (TCGA): 100/497 (20.1%)

Cancer IHC. lung cancer: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. Inhibins and activins inhibit and activate, respectively, the secretion of follitropin by the pituitary gland. Inhibins/activins are involved in regulating a number of diverse functions such as hypothalamic and pituitary hormone secretion, gonadal hormone secretion, germ cell development and maturation, erythroid differentiation, insulin secretion, nerve cell survival, embryonic axial development or bone growth, depending on their ... [full text in table]

Curated tissue note. Originally found in ovary (granulosa cells) and testis (Sertoli cells), but widely distributed in many tissues including brain and placenta. In adrenal cortex expression is limited to the zona reticularis and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P05111 | HPA portal (live; not the pinned values)

INO80B | Unsafe / RNA

ENSG00000115274

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 25.5 nTPM (limit 10); adrenal gland: 29.5 nTPM (limit 2); amygdala: 26.7 nTPM (limit 2); basal ganglia: 26.2 nTPM (limit 2); bone marrow: 20.3 nTPM (limit 2); cerebellum: 36.4 nTPM (limit 2); cerebral cortex: 29 nTPM (limit 2); choroid plexus: 15.5 nTPM (limit 2); colon: 23.3 nTPM ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Induces growth and cell cycle arrests at the G1 phase of the cell cycle Proposed core component of the chromatin remodeling INO80 complex which is involved in transcriptional regulation, DNA replication and probably DNA repair

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9C086 | HPA portal (live; not the pinned values)

INSL3 | Unsafe / High RNA

ENSG00000248099

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 11.8 nTPM; testis/macrophages; hpa_single_cell_tissue; vital; 43.73 TPM; blood/TFH; dice_tfh; vital

Cancer RNA. Colon Adenocarcinoma (validation): 207/486 (42.6%); Rectum Adenocarcinoma (validation): 84/207 (40.6%); Testicular Germ Cell Tumor (TCGA): 36/133 (27.1%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Seems to play a role in testicular function. May be a trophic hormone with a role in testicular descent in fetal life. Is a ligand for LGR8 receptor

Curated tissue note. Expressed in prenatal and postnatal Leydig cells. Found as well in the corpus luteum, trophoblast, fetal membranes and breast

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P51460 | HPA portal (live; not the pinned values)

INSL6 | Unsafe / RNA

ENSG00000120210

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 6.4 nTPM (limit 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 17/349 (4.9%); Lung Squamous Cell Carcinoma (validation): 3/68 (4.4%); Stomach Adenocarcinoma (TCGA): 11/346 (3.2%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May have a role in sperm development and fertilization

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y581 | HPA portal (live; not the pinned values)

INSRR | Unsafe / RNA

ENSG00000027644

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 3.7 nTPM (limit 2); kidney: 3.7 nTPM (baseline 2)

Cancer RNA. Kidney Chromophobe (TCGA): 4/64 (6.2%); Glioblastoma Multiforme (validation): 3/58 (5.2%); Glioblastoma Multiforme (TCGA): 2/141 (1.4%)

Cancer IHC. cervical cancer: 12/12 (100.0%); stomach cancer: 12/12 (100.0%)

Biology. Receptor with tyrosine-protein kinase activity. Functions as a pH sensing receptor which is activated by increased extracellular pH. Activates an intracellular signaling pathway that involves IRS1 and AKT1/PKB

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P14616 | HPA portal (live; not the pinned values)

IPO11 | Unsafe / Protein

ENSG00000086200

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: Medium (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / goblet cells: Medium (Approved); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. prostate cancer: 8/11 (72.7%); liver cancer: 6/11 (54.5%)

Biology. Functions in nuclear protein import as nuclear transport receptor. Serves as receptor for nuclear localization signals (NLS) in cargo substrates. Is thought to mediate docking of the importin/substrate complex to the nuclear pore complex (NPC) through binding to nucleoporin and the complex is subsequently translocated through the pore by an energy requiring, Ran-dependent mechanism. At the nucleoplasmic side of the NPC, Ran binds to ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UI26 | HPA portal (live; not the pinned values)

IQCD | Unsafe / RNA

ENSG00000166578

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.2 nTPM (limit 2); basal ganglia: 3.2 nTPM (limit 2); choroid plexus: 21.4 nTPM (limit 2); hippocampal formation: 2.6 nTPM (limit 2); hypothalamus: 3 nTPM (limit 2); midbrain: 2.2 nTPM (limit 2); pituitary gland: 3.8 nTPM (limit 2); retina: 3.6 nTPM (limit 2); spinal cord: 2.5 nTPM ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 44/50 (88.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 209/282 (74.1%); Uterine Corpus Endometrial Carcinoma (TCGA): 108/176 (61.4%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Component of the nexin-dynein regulatory complex (N-DRC), a key regulator of ciliary/flagellar motility which maintains the alignment and integrity of the distal axoneme and regulates microtubule sliding in motile axonemes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96DY2 | HPA portal (live; not the pinned values)

IQCH | Unsafe / RNA

ENSG00000103599

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 2.1 nTPM (limit 2); retina: 4.8 nTPM (limit 2); kidney: 2.1 nTPM (baseline 2); retina: 4.8 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 39/50 (78.0%); Glioblastoma Multiforme (validation): 26/58 (44.8%); Pancreatic Adenocarcinoma (validation): 15/80 (18.8%)

Cancer IHC. breast cancer: 12/12 (100.0%); liver cancer: 12/12 (100.0%)

Biology. May play a regulatory role in spermatogenesis

Curated tissue note. Expressed in fetal and adult testis, in germ cells but not somatic cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86VS3 | HPA portal (live; not the pinned values)

IQCN | Unsafe / Protein

ENSG00000130518

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebellum / Bergmann glia - nucleus: High (Enhanced); cerebellum / GLUC cells - nucleus: Low (Enhanced); cerebellum / Purkinje cells - nucleus: High (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (validation): 81/207 (39.1%); Colon Adenocarcinoma (validation): 148/486 (30.5%); Ovary Serous Cystadenocarcinoma (validation): 24/81 (29.6%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Essential for spermiogenesis and fertilization (PubMed:36321563). May be required for manchette assembly in elongating spermatids (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0B3 | HPA portal (live; not the pinned values)

IQUB | Unsafe / RNA

ENSG00000164675

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.3 nTPM (limit 2); kidney: 2.9 nTPM (limit 2); choroid plexus: 2.3 nTPM (baseline 2); kidney: 2.9 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 27/58 (46.6%); Breast Invasive Carcinoma (validation): 18/50 (36.0%); Kidney Renal Clear Cell Carcinoma (validation): 14/100 (14.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Adapter protein that anchors the radial spoke 1 (RS1) complex to the A microtubule of outer doublet microtubules in axonemes (PubMed:36355624). The triple radial spokes (RS1, RS2 and RS3) are required to modulate beating of the sperm flagellum (PubMed:36355624). May play a role in inhibiting signaling via MAPK1/ERK2 and MAPK3/ERK1 (PubMed:36355624). Additionally, may play a role in the functioning of cilia (By similarity). Not required ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NA54 | HPA portal (live; not the pinned values)

IRAK1BP1 | Unsafe / RNA

ENSG00000146243

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 5.3 nTPM (limit 2); kidney: 2.8 nTPM (limit 2); lung: 2.1 nTPM (limit 2); pancreas: 3.1 nTPM (limit 2); pituitary gland: 3.3 nTPM (limit 2); retina: 5.4 nTPM (limit 2); bone marrow: 5.3 nTPM (baseline 2); kidney: 2.8 nTPM (baseline 2); lung: 2.1 nTPM (baseline 2); pancreas: 3.1 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 47/50 (94.0%); Rectum Adenocarcinoma (validation): 147/207 (71.0%)

Cancer IHC. colorectal cancer: 10/11 (90.9%); renal cancer: 10/12 (83.3%)

Biology. Component of the IRAK1-dependent TNFRSF1A signaling pathway that leads to NF-kappa-B activation and is required for cell survival. Acts by enhancing RELA transcriptional activity (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VVH5 | HPA portal (live; not the pinned values)

ISM2 | Unsafe / Protein

ENSG00000100593

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / endothelial cells: Low (Supported); colon / endothelial cells: Low (Supported)

Cancer RNA. Kidney Chromophobe (TCGA): 27/64 (42.2%); Testicular Germ Cell Tumor (TCGA): 19/133 (14.3%); Rectum Adenocarcinoma (TCGA): 10/88 (11.4%)

Cancer IHC. colorectal cancer: 3/10 (30.0%); pancreatic cancer: 1/11 (9.1%)

Biology. Expressed at high levels in the placenta and at moderate levels in the pancreas, kidney, heart, liver, lung, brain and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6H9L7 | HPA portal (live; not the pinned values)

ISY1 | Unsafe / Protein

ENSG00000240682

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / glial cells: High (Supported); caudate / neuronal cells: High (Supported); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Component of the spliceosome C complex required for the selective processing of microRNAs during embryonic stem cell differentiation (By similarity). Required for the biogenesis of all miRNAs from the pri-miR-17-92 primary transcript except miR-92a (By similarity). Only required for the biogenesis of miR-290 and miR-96 from the pri-miR-290-295 and pri-miR-96-183 primary transcripts, respectively (By similarity). Required during the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9ULR0 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ITIH4 | Unsafe / RNA

ENSG00000055955

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 17.9 nTPM (limit 12); basal ganglia: 2.7 nTPM (limit 2); choroid plexus: 2.7 nTPM (limit 2); liver: 1807.4 nTPM (limit 12); pancreas: 47.5 nTPM (limit 12); pituitary gland: 4.2 nTPM (limit 2); retina: 3.2 nTPM (limit 2); skeletal muscle: 38.8 nTPM (limit 12); adrenal gland: 17.9 nTPM ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 228/231 (98.7%); Liver Hepatocellular Carcinoma (TCGA): 356/362 (98.3%); Breast Invasive Carcinoma (validation): 42/50 (84.0%)

Cancer IHC. liver cancer: 5/12 (41.7%); breast cancer: 2/12 (16.7%)

Biology. Type II acute-phase protein (APP) involved in inflammatory responses to trauma. May also play a role in liver development or regeneration

Curated tissue note. Liver specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14624 | HPA portal (live; not the pinned values)

ITPRIPL1 | Unsafe / RNA

ENSG00000198885

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 3.2 nTPM (limit 2); salivary gland: 7.3 nTPM (limit 6); cerebellum: 3.2 nTPM (baseline 2); salivary gland: 7.3 nTPM (baseline 6)

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Glioblastoma Multiforme (TCGA): 94/141 (66.7%); Testicular Germ Cell Tumor (TCGA): 40/133 (30.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Functions as a ligand of CD3E, inhibiting TCR-CD3 complex signaling to regulate T cell activation. Induces stable CD3E-NCK1 binding, thereby preventing the CD3E-ZAP70 interaction and subsequently inhibiting the activation of the downstream ERK-NFkB signaling cascade and calcium influx

Curated tissue note. Expressed in testis and tumoral cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6GPH6 | HPA portal (live; not the pinned values)

IZUMO4 | Unsafe / Protein

ENSG00000099840

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / proximal tubules (cell body): Medium (Enhanced)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 293/480 (61.0%); Glioblastoma Multiforme (TCGA): 83/141 (58.9%); Glioblastoma Multiforme (validation): 15/58 (25.9%)

Cancer IHC. testis cancer: 11/12 (91.7%); glioma: 4/11 (36.4%)

Biology. Detected in sperm

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q1ZYL8 | HPA portal (live; not the pinned values)

JAM3 | Unsafe / Protein

ENSG00000166086

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bronchus / ciliated cells (cilia axoneme): High (Approved); bronchus / ciliated cells (tip of cilia): High (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / neuropil: Medium (Approved); colon / endothelial cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 78/80 (97.5%)

Cancer IHC. glioma: 11/11 (100.0%); testis cancer: 8/12 (66.7%)

Biology. Junctional adhesion protein that mediates heterotypic cell-cell interactions with its cognate receptor JAM2 to regulate different cellular processes (PubMed:11590146, PubMed:11823489). Plays a role in homing and mobilization of hematopoietic stem and progenitor cells within the bone marrow. At the surface of bone marrow stromal cells, it contributes to the retention of the hematopoietic stem and progenitor cells expressing JAM3 ... [full text in table]

Curated tissue note. Detected on round and elongated spermatids (at protein level) (PubMed:15372036). Highest expression in placenta, brain and kidney. Significant expression is detected on platelets. Expressed in intestinal ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BX67 | HPA portal (live; not the pinned values)

JMJD1C | Unsafe / Protein

ENSG00000171988

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. testis cancer: 12/12 (100.0%); urothelial cancer: 12/12 (100.0%)

Biology. Demethylates lysine in proteins, such as STAT3 or MDC1 (By similarity) (PubMed:24240613). Demethylates MDC1, thereby promoting MDC1-RNF8 interaction and facilitating RNF8-dependent MDC1 ubiquitination essential for double-strand break (DSB) repair (PubMed:24240613). Demethylation of STAT3 at 'Lys-140' facilitates its interactions with the phosphatase PTPN6 and restrains STAT3 activation (By similarity). It is uncertain whether JMJD1C ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15652 | HPA portal (live; not the pinned values)

JMY | Unsafe / Protein

ENSG00000152409

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / Purkinje cells: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 476/486 (97.9%); Pancreatic Adenocarcinoma (validation): 76/80 (95.0%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); cervical cancer: 10/11 (90.9%)

Biology. Acts both as a nuclear p53/TP53-cofactor and a cytoplasmic regulator of actin dynamics depending on conditions (PubMed:30420355). In nucleus, acts as a cofactor that increases p53/TP53 response via its interaction with p300/EP300. Increases p53/TP53-dependent transcription and apoptosis, suggesting an important role in p53/TP53 stress response such as DNA damage. In cytoplasm, acts as a nucleation-promoting factor for both branched and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N9B5 | HPA portal (live; not the pinned values)

KANSL1L | Unsafe / Protein

ENSG00000144445

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / cells in ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%)

Cancer IHC. colorectal cancer: 5/12 (41.7%); pancreatic cancer: 4/12 (33.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0AUZ9 | HPA portal (live; not the pinned values)

KANSL3 | Unsafe / Protein

ENSG00000114982

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: Medium (Approved); bronchus / ciliated cells (cell body): Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 97/100 (97.0%)

Cancer IHC. prostate cancer: 11/12 (91.7%); liver cancer: 9/11 (81.8%)

Biology. Non-catalytic component of the NSL histone acetyltransferase complex, a multiprotein complex that mediates histone H4 acetylation at 'Lys-5'- and 'Lys-8' (H4K5ac and H4K8ac) at transcription start sites and promotes transcription initiation (PubMed:20018852, PubMed:33657400). The NSL complex also acts as a regulator of gene expression in mitochondria (PubMed:27768893). Within the NSL complex, KANSL3 is required to promote KAT8 ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P2N6 | HPA portal (live; not the pinned values)

KASH5 | Unsafe / Protein

ENSG00000161609

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / cells in granular layer: Medium (Enhanced); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 13/133 (9.8%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 11/283 (3.9%); Uterine Corpus Endometrial Carcinoma (TCGA): 3/176 (1.7%)

Cancer IHC. ovarian cancer: 5/12 (41.7%); renal cancer: 5/12 (41.7%)

Biology. As a component of the LINC (LInker of Nucleoskeleton and Cytoskeleton) complex, involved in the connection between the nuclear lamina and the cytoskeleton. The nucleocytoplasmic interactions established by the LINC complex play an important role in the transmission of mechanical forces across the nuclear envelope and in nuclear movement and positioning. Required for telomere attachment to nuclear envelope in the prophase of meiosis ... [full text in table]

Curated tissue note. Expressed in testis (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N6L0 | HPA portal (live; not the pinned values)

KATNAL1 | Unsafe / RNA

ENSG00000102781

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.7 nTPM (limit 2); amygdala: 4.7 nTPM (limit 2); basal ganglia: 4.3 nTPM (limit 2); bone marrow: 6.4 nTPM (limit 2); cerebellum: 5.8 nTPM (limit 2); cerebral cortex: 9.6 nTPM (limit 2); choroid plexus: 2.5 nTPM (limit 2); colon: 8.4 nTPM (limit 6); heart muscle: 17.3 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Pancreatic Adenocarcinoma (validation): 70/80 (87.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Regulates microtubule dynamics in Sertoli cells, a process that is essential for spermiogenesis and male fertility. Severs microtubules in an ATP-dependent manner, promoting rapid reorganization of cellular microtubule arrays (By similarity). Has microtubule-severing activity in vitro (PubMed:26929214)

Curated tissue note. Expressed in testis, restricted to Sertoli cells (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BW62 | HPA portal (live; not the pinned values)

KBTBD8 | Unsafe / Protein

ENSG00000163376

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / glial cells: Low (Approved); cerebellum / cells in granular layer: Medium (Approved); colon / glandular cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Breast Invasive Carcinoma (validation): 15/50 (30.0%); Rectum Adenocarcinoma (validation): 52/207 (25.1%)

Cancer IHC. lymphoma: 7/11 (63.6%); stomach cancer: 7/11 (63.6%)

Biology. Substrate-specific adapter of a BCR (BTB-CUL3-RBX1) E3 ubiquitin ligase complex that acts as a regulator of neural crest specification (PubMed:26399832). The BCR(KBTBD8) complex acts by mediating monoubiquitination of NOLC1 and TCOF1: monoubiquitination promotes the formation of a NOLC1-TCOF1 complex that acts as a platform to connect RNA polymerase I with enzymes responsible for ribosomal processing and modification, leading to ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NFY9 | HPA portal (live; not the pinned values)

KCNA3 | Unsafe / Protein

ENSG00000177272

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebral cortex / glial cells: Medium (Approved); hippocampus / glial cells: Medium (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 31/58 (53.4%); Rectum Adenocarcinoma (validation): 108/207 (52.2%); Lung Adenocarcinoma (validation): 51/105 (48.6%)

Cancer IHC. glioma: 4/11 (36.4%); cervical cancer: 1/12 (8.3%)

Biology. Mediates the voltage-dependent potassium ion permeability of excitable membranes. Assuming opened or closed conformations in response to the voltage difference across the membrane, the protein forms a potassium-selective channel through which potassium ions may pass in accordance with their electrochemical gradient Lacks voltage-gated potassium channel activity

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P22001 | HPA portal (live; not the pinned values)

KCNH5 | Unsafe / RNA

ENSG00000140015

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.3 nTPM (limit 2); retina: 4.3 nTPM (limit 2); cerebral cortex: 2.3 nTPM (baseline 2); retina: 4.3 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 6/58 (10.3%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%)

Cancer IHC. lung cancer: 2/11 (18.2%); ovarian cancer: 2/11 (18.2%)

Biology. Pore-forming (alpha) subunit of a voltage-gated delayed rectifier potassium channel that mediates outward-rectifying potassium currents which, on depolarization, reaches a steady-state level and do not inactivate (PubMed:11943152, PubMed:12135768, PubMed:24133262, PubMed:36928654). The kinetic is characterized by a slow activation time course and a small voltage dependence of the activation time constants, therefore, starts to open at ... [full text in table]

Curated tissue note. Detected in brain, skeletal muscle, heart, placenta, lung and liver, and at low levels in kidney

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCM2 | HPA portal (live; not the pinned values)

KCNJ14 | Unsafe / RNA

ENSG00000182324

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 4.5 nTPM (limit 2); retina: 56.9 nTPM (limit 2); basal ganglia: 4.5 nTPM (baseline 2); retina: 56.9 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 16/50 (32.0%); Glioblastoma Multiforme (validation): 16/58 (27.6%); Uterine Corpus Endometrial Carcinoma (TCGA): 18/176 (10.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Inward rectifier potassium channels are characterized by a greater tendency to allow potassium to flow into the cell rather than out of it. Their voltage dependence is regulated by the concentration of extracellular potassium; as external potassium is raised, the voltage range of the channel opening shifts to more positive voltages

Curated tissue note. Expressed preferentially in retina

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UNX9 | HPA portal (live; not the pinned values)

KCNK13 | Unsafe / Protein

ENSG00000152315

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); hippocampus / glial cells: Low (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 28/58 (48.3%); Testicular Germ Cell Tumor (TCGA): 64/133 (48.1%); Thyroid Carcinoma (TCGA): 185/495 (37.4%)

Cancer IHC. glioma: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. K(+) channel that conducts outward rectifying tonic currents potentiated by purinergic signals (PubMed:24163367, PubMed:25148687, PubMed:30472253, PubMed:38409076). Homo- and heterodimerizes to form functional channels with distinct regulatory and gating properties (PubMed:25148687, PubMed:40011745, PubMed:40011746, PubMed:40307591, PubMed:40178898). Contributes most of K(+) currents at the plasma membrane of resting microglia ... [full text in table]

Curated tissue note. Expressed in microglia (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HB14 | HPA portal (live; not the pinned values)

KCNMB2 | Unsafe / Protein

ENSG00000197584

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); cerebellum / cells in molecular layer: Low (Uncertain); cerebral cortex / glial cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 24/58 (41.4%); Pancreatic Adenocarcinoma (TCGA): 37/176 (21.0%); Ovary Serous Cystadenocarcinoma (TCGA): 73/349 (20.9%)

Cancer IHC. prostate cancer: 8/12 (66.7%); breast cancer: 6/12 (50.0%)

Biology. Regulatory subunit of the calcium activated potassium KCNMA1 (maxiK) channel. Modulates the calcium sensitivity and gating kinetics of KCNMA1, thereby contributing to KCNMA1 channel diversity. Acts as a negative regulator that confers rapid and complete inactivation of KCNMA1 channel complex. May participate in KCNMA1 inactivation in chromaffin cells of the adrenal gland or in hippocampal CA1 neurons

Curated tissue note. Expressed in kidney, heart and brain. Highly expressed in ovary. Expressed at low level in other tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y691 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

KCNMB3 | Unsafe / Protein

ENSG00000171121

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: High (Approved); duodenum / glandular cells: High (Approved); esophagus / squamous epithelial cells: Low (Approved); heart muscle / cardiomyocytes: Low (Approved); kidney / cells ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Liver Hepatocellular Carcinoma (validation): 84/231 (36.4%); Pancreatic Adenocarcinoma (validation): 17/80 (21.2%)

Cancer IHC. colorectal cancer: 8/11 (72.7%); stomach cancer: 7/10 (70.0%)

Biology. Regulatory subunit of the calcium activated potassium KCNMA1 (maxiK) channel. Modulates the calcium sensitivity and gating kinetics of KCNMA1, thereby contributing to KCNMA1 channel diversity. Alters the functional properties of the current expressed by the KCNMA1 channel. Isoform 2, isoform 3 and isoform 4 partially inactivate the current of KCNBMA. Isoform 4 induces a fast and incomplete inactivation of KCNMA1 channel that is ... [full text in table]

Curated tissue note. Isoform 1, isoform 3 and isoform 4 are widely expressed. Isoform 2 is expressed placenta, pancreas, kidney and heart. Isoform 1 and isoform 3 are highly expressed in pancreas and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NPA1 | HPA portal (live; not the pinned values)

KCNV2 | Unsafe / Protein

ENSG00000168263

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: Medium (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 62/133 (46.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Ovary Serous Cystadenocarcinoma (TCGA): 1/349 (0.3%)

Cancer IHC. pancreatic cancer: 1/10 (10.0%); breast cancer: 0/8 (0.0%)

Biology. Potassium channel subunit. Modulates channel activity by shifting the threshold and the half-maximal activation to more negative values

Curated tissue note. Detected in lung, liver, kidney, pancreas, spleen, thymus, prostate, testis, ovary and colon

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TDN2 | HPA portal (live; not the pinned values)

KCTD19 | Unsafe / RNA

ENSG00000168676

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 3.3 nTPM (limit 2); bone marrow: 3.3 nTPM (baseline 2)

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 27/283 (9.5%); Testicular Germ Cell Tumor (TCGA): 12/133 (9.0%); Pancreatic Adenocarcinoma (validation): 5/80 (6.2%)

Cancer IHC. pancreatic cancer: 4/10 (40.0%); melanoma: 4/11 (36.4%)

Biology. Transcription regulator which is essential for male fertility and for the completion of meiotic prophase in spermatocytes. Regulates progression of the pachytene stage of meiotic prophase and promotes the transcriptional activation activity ZNF541. Required for the organization of chromosomes during metaphase I

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q17RG1 | HPA portal (live; not the pinned values)

KDM4C | Unsafe / RNA

ENSG00000107077

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 9.3 nTPM (limit 2); amygdala: 6.8 nTPM (limit 2); basal ganglia: 7.6 nTPM (limit 2); bone marrow: 29.7 nTPM (limit 2); cerebellum: 38.3 nTPM (limit 2); cerebral cortex: 7.6 nTPM (limit 2); choroid plexus: 16.3 nTPM (limit 2); colon: 13.3 nTPM (limit 6); duodenum: 12.7 nTPM (limit 2); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Histone demethylase that specifically demethylates 'Lys-9' and 'Lys-36' residues of histone H3, thereby playing a central role in histone code. Does not demethylate histone H3 'Lys-4', H3 'Lys-27' nor H4 'Lys-20'. Demethylates trimethylated H3 'Lys-9' and H3 'Lys-36' residue, while it has no activity on mono- and dimethylated residues. Demethylation of Lys residue generates formaldehyde and succinate

Curated tissue note. Overexpressed in several esophageal squamous cell carcinomas (ESCs)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H3R0 | HPA portal (live; not the pinned values)

KDM4D | Unsafe / Protein

ENSG00000186280

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); colon / glandular cells: Low (Approved); duodenum / glandular cells: Low (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 32/58 (55.2%); Testicular Germ Cell Tumor (TCGA): 14/133 (10.5%); Breast Invasive Carcinoma (validation): 4/50 (8.0%)

Cancer IHC. cervical cancer: 2/12 (16.7%); lung cancer: 1/9 (11.1%)

Biology. Histone demethylase that specifically demethylates 'Lys-9' of histone H3, thereby playing a central role in histone code. Does not demethylate histone H3 'Lys-4', H3 'Lys-27', H3 'Lys-36' nor H4 'Lys-20'. Demethylates both di- and trimethylated H3 'Lys-9' residue, while it has no activity on monomethylated residues. Demethylation of Lys residue generates formaldehyde and succinate

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6B0I6 | HPA portal (live; not the pinned values)

KDM7A | Unsafe / Protein

ENSG00000006459

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); skin cancer: 12/12 (100.0%)

Biology. Histone demethylase required for brain development. Specifically demethylates dimethylated 'Lys-9', 'Lys-27' and 'Lys-36' (H3K9me2, H3K27me2, H3K36me2, respectively) of histone H3 and monomethylated histone H4 'Lys-20' residue (H4K20Me1), thereby playing a central role in histone code (PubMed:20023638, PubMed:20622853). Specifically binds trimethylated 'Lys-4' of histone H3 (H3K4me3), affecting histone demethylase specificity: in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZMT4 | HPA portal (live; not the pinned values)

KHDC1 | Unsafe / RNA

ENSG00000135314

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 11 nTPM (limit 2); basal ganglia: 14.2 nTPM (limit 2); cerebellum: 9.2 nTPM (limit 2); cerebral cortex: 13.7 nTPM (limit 2); choroid plexus: 4 nTPM (limit 2); heart muscle: 3.7 nTPM (limit 2); hippocampal formation: 9.2 nTPM (limit 2); hypothalamus: 9.4 nTPM (limit 2); midbrain: 9.9 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 135/141 (95.7%); Glioblastoma Multiforme (validation): 36/58 (62.1%); Breast Invasive Carcinoma (validation): 29/50 (58.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4VXA5 | HPA portal (live; not the pinned values)

KHDC1L | Unsafe / RNA

ENSG00000256980

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 8.1 nTPM (limit 2); basal ganglia: 8.1 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 92/133 (69.2%); Head and Neck Squamous Cell Carcinoma (TCGA): 279/492 (56.7%); Lung Squamous Cell Carcinoma (TCGA): 93/489 (19.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JSQ8 | HPA portal (live; not the pinned values)

KHDRBS3 | Unsafe / Protein

ENSG00000131773

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Enhanced); adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Medium (Enhanced); caudate / neuronal cells: High (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 266/282 (94.3%)

Cancer IHC. glioma: 11/11 (100.0%); colorectal cancer: 10/11 (90.9%)

Biology. RNA-binding protein that plays a role in the regulation of alternative splicing and influences mRNA splice site selection and exon inclusion. Binds preferentially to the 5'-[AU]UAAA-3' motif in vitro. Binds optimally to RNA containing 5'-[AU]UAA-3' as a bipartite motif spaced by more than 15 nucleotides. Binds poly(A). RNA-binding abilities are down-regulated by tyrosine kinase PTK6 (PubMed:10564820, PubMed:19561594, PubMed:26758068). ... [full text in table]

Curated tissue note. Ubiquitous with higher expression in testis, skeletal muscle and brain. Expressed in the kidney only in podocytes, the glomerular epithelial cells of the kidney. Strongly expressed after meiosis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75525 | HPA portal (live; not the pinned values)

KIAA0825 | Unsafe / Protein

ENSG00000185261

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / Purkinje cells: Low (Uncertain); cerebral cortex / neuronal cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 51/58 (87.9%); Pancreatic Adenocarcinoma (validation): 29/80 (36.2%); Kidney Renal Papillary Cell Carcinoma (TCGA): 5/282 (1.8%)

Cancer IHC. liver cancer: 7/10 (70.0%); pancreatic cancer: 4/9 (44.4%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IV33 | HPA portal (live; not the pinned values)

KIAA1328 | Unsafe / Protein

ENSG00000150477

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); breast cancer: 11/12 (91.7%)

Biology. Competes with SMC1 for binding to SMC3. May affect the availability of SMC3 to engage in the formation of multimeric protein complexes

Curated tissue note. Widely expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86T90 | HPA portal (live; not the pinned values)

KIAA1549 | Unsafe / Protein

ENSG00000122778

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Rectum Adenocarcinoma (validation): 191/207 (92.3%); Breast Invasive Carcinoma (validation): 46/50 (92.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. May play a role in photoreceptor function

Curated tissue note. Expression is low to moderate in retina and tissues, such as heart and kidney, and is predominantly expressed in brain Abundantly expressed in the retina, compared with only minimal expression in brain and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HCM3 | HPA portal (live; not the pinned values)

KIAA1586 | Unsafe / Protein

ENSG00000168116

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Approved); cerebral cortex / neuropil: Low (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: Medium (Approved); colon / peripheral nerve/ganglion: Medium (Approved); duodenum / glandular cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Lung Squamous Cell Carcinoma (validation): 56/68 (82.4%)

Cancer IHC. urothelial cancer: 5/8 (62.5%); pancreatic cancer: 6/10 (60.0%)

Biology. E3 SUMO-protein ligase; facilitates UBE2I/UBC9-mediated SUMO2 modification of target proteins (PubMed:26524493)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HCI6 | HPA portal (live; not the pinned values)

KICS2 | Unsafe / Protein

ENSG00000174206

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Uncertain); cerebral cortex / neuropil: Low (Uncertain); colon / endothelial cells: Low (Uncertain); colon / glandular cells: High (Uncertain); colon / peripheral nerve/ganglion: Medium (Uncertain); duodenum / glandular cells: Medium (Uncertain); kidney / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 205/207 (99.0%); Colon Adenocarcinoma (validation): 476/486 (97.9%)

Cancer IHC. cervical cancer: 2/11 (18.2%); ovarian cancer: 2/11 (18.2%)

Biology. As part of the KICSTOR complex functions in the amino acid-sensing branch of the TORC1 signaling pathway. Recruits, in an amino acid-independent manner, the GATOR1 complex to the lysosomal membranes and allows its interaction with GATOR2 and the RAG GTPases. Functions upstream of the RAG GTPases and is required to negatively regulate mTORC1 signaling in absence of amino acids. In absence of the KICSTOR complex mTORC1 is constitutively ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96MD2 | HPA portal (live; not the pinned values)

KIF14 | Unsafe / RNA

ENSG00000118193

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 6.5 nTPM (limit 2); thymus: 10.6 nTPM (limit 10); bone marrow: 6.5 nTPM (baseline 2); thymus: 10.6 nTPM (baseline 10)

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Colon Adenocarcinoma (validation): 409/486 (84.2%); Rectum Adenocarcinoma (validation): 149/207 (72.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); ovarian cancer: 12/12 (100.0%)

Biology. Microtubule motor protein that binds to microtubules with high affinity through each tubulin heterodimer and has an ATPase activity (By similarity). Plays a role in many processes like cell division, cytokinesis and also in cell proliferation and apoptosis (PubMed:16648480, PubMed:24784001). During cytokinesis, targets to central spindle and midbody through its interaction with PRC1 and CIT respectively (PubMed:16431929). Regulates ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15058 | HPA portal (live; not the pinned values)

KIF15 | Unsafe / Protein

ENSG00000163808

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebral cortex / neuropil: Low (Approved); colon / glandular cells: High (Approved); duodenum / glandular cells: High (Approved); heart ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 41/50 (82.0%); Colon Adenocarcinoma (validation): 297/486 (61.1%)

Cancer IHC. lymphoma: 8/12 (66.7%); cervical cancer: 6/11 (54.5%)

Biology. Plus-end directed kinesin-like motor enzyme involved in mitotic spindle assembly

Curated tissue note. Expressed in testis, colon, thymus and in breast cancer

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NS87 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

KIF18A | Unsafe / Protein

ENSG00000121621

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Colon Adenocarcinoma (validation): 415/486 (85.4%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Microtubule-depolymerizing kinesin which plays a role in chromosome congression by reducing the amplitude of preanaphase oscillations and slowing poleward movement during anaphase, thus suppressing chromosome movements. May stabilize the CENPE-BUB1B complex at the kinetochores during early mitosis and maintains CENPE levels at kinetochores during chromosome congression

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NI77 | HPA portal (live; not the pinned values)

KIF20A | Unsafe / Protein

ENSG00000112984

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. 42.7 nTPM; placenta/mixed immune cells; hpa_single_cell_tissue; vital; 42.95 TPM; blood/NK-cell; hpa_immune_cell_samples; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 132/133 (99.2%); Colon Adenocarcinoma (validation): 480/486 (98.8%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 279/283 (98.6%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); stomach cancer: 11/11 (100.0%)

Biology. Mitotic kinesin required for chromosome passenger complex (CPC)-mediated cytokinesis. Following phosphorylation by PLK1, involved in recruitment of PLK1 to the central spindle. Interacts with guanosine triphosphate (GTP)-bound forms of RAB6A and RAB6B. May act as a motor required for the retrograde RAB6 regulated transport of Golgi membranes and associated vesicles along microtubules. Has a microtubule plus end-directed motility

Therapeutic evidence. Trial registration only.

UniProt O95235 | Trial/source record | HPA portal (live; not the pinned values)

KIF20B | Unsafe / Protein

ENSG00000138182

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. 198.9 nTPM; tongue/basal keratinocytes; hpa_single_cell_tissue; protected; 59.5 TPM; blood/CD4_STIM; dice_cd4_stim; vital

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 483/486 (99.4%); Rectum Adenocarcinoma (validation): 204/207 (98.6%)

Cancer IHC. colorectal cancer: 4/12 (33.3%); ovarian cancer: 4/12 (33.3%)

Biology. Plus-end-directed motor enzyme that is required for completion of cytokinesis (PubMed:11470801, PubMed:12740395). Required for proper midbody organization and abscission in polarized cortical stem cells. Plays a role in the regulation of neuronal polarization by mediating the transport of specific cargos. Participates in the mobilization of SHTN1 and in the accumulation of PIP3 in the growth cone of primary hippocampal neurons in a ... [full text in table]

Curated tissue note. Brain, ovary, kidney and testis (at protein level) (PubMed:12740395). Overexpressed in bladder cancer cells (at protein level) (PubMed:17409436). Expressed in testis. Overexpressed in bladder cancer cells ... [full text in table]

Therapeutic evidence. Trial registration only.

UniProt Q96Q89 | Trial/source record | HPA portal (live; not the pinned values)

KIF24 | Unsafe / RNA

ENSG00000186638

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.1 nTPM (limit 2); choroid plexus: 3.5 nTPM (limit 2); bone marrow: 2.1 nTPM (baseline 2); choroid plexus: 3.5 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 49/58 (84.5%); Breast Invasive Carcinoma (validation): 37/50 (74.0%); Colon Adenocarcinoma (validation): 325/486 (66.9%)

Cancer IHC. melanoma: 5/11 (45.5%); testis cancer: 4/9 (44.4%)

Biology. Microtubule-dependent motor protein that acts as a negative regulator of ciliogenesis by mediating recruitment of CCP110 to mother centriole in cycling cells, leading to restrict nucleation of cilia at centrioles. Mediates depolymerization of microtubules of centriolar origin, possibly to suppress aberrant cilia formation (PubMed:21620453). Following activation by NEK2 involved in disassembly of primary cilium during G2/M phase but ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T7B8 | HPA portal (live; not the pinned values)

KIF2A | Unsafe / Protein

ENSG00000068796

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: Low (Approved); cerebellum / ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. lymphoma: 11/12 (91.7%); glioma: 10/11 (90.9%)

Biology. Plus end-directed microtubule-dependent motor required for normal brain development. May regulate microtubule dynamics during axonal growth. Required for normal progression through mitosis. Required for normal congress of chromosomes at the metaphase plate. Required for normal spindle dynamics during mitosis. Promotes spindle turnover. Implicated in formation of bipolar mitotic spindles. Has microtubule depolymerization activity

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00139 | HPA portal (live; not the pinned values)

KIF2B | Unsafe / Protein

ENSG00000141200

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. renal cancer: 7/11 (63.6%); ovarian cancer: 6/12 (50.0%)

Biology. Plus end-directed microtubule-dependent motor required for spindle assembly and chromosome movement. Has microtubule depolymerization activity (PubMed:17538014). Plays a role in chromosome congression (PubMed:23891108)

Curated tissue note. Highest level in lung. High level in ovary, moderate levels in heart, kidney, placenta, skeletal muscle and spleen (at protein level). Pancreas and spleen express a shorter isoform (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N4N8 | HPA portal (live; not the pinned values)

KIF2C | Unsafe / Protein

ENSG00000142945

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); duodenum / endocrine cells: Low (Enhanced); lymph node / germinal center cells: Low (Enhanced); rectum / endocrine cells: Low (Enhanced); small intestine / enterocytes: Low (Enhanced); thymus / cortical cells: Medium (Enhanced); thymus / medullary ... [full text in table]

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 283/283 (100.0%); Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%); Colon Adenocarcinoma (validation): 484/486 (99.6%)

Cancer IHC. cervical cancer: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. In complex with KIF18B, constitutes the major microtubule plus-end depolymerizing activity in mitotic cells (PubMed:21820309). Regulates the turnover of microtubules at the kinetochore and functions in chromosome segregation during mitosis (PubMed:19060894). Plays a role in chromosome congression and is required for the lateral to end-on conversion of the chromosome-microtubule attachment (PubMed:23891108)

Curated tissue note. Expressed at high levels in thymus and testis, at low levels in small intestine, the mucosal lining of colon, and placenta, and at very low levels in spleen and ovary; expression is not detected in prostate, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q99661 | HPA portal (live; not the pinned values)

KIF4A | Unsafe / Protein

ENSG00000090889

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Colon Adenocarcinoma (validation): 476/486 (97.9%); Colon Adenocarcinoma (TCGA): 244/254 (96.1%)

Cancer IHC. prostate cancer: 12/12 (100.0%); breast cancer: 11/11 (100.0%)

Biology. Iron-sulfur (Fe-S) cluster binding motor protein that has a role in chromosome segregation during mitosis (PubMed:29848660). Translocates PRC1 to the plus ends of interdigitating spindle microtubules during the metaphase to anaphase transition, an essential step for the formation of an organized central spindle midzone and midbody and for successful cytokinesis (PubMed:15297875, PubMed:15625105). May play a role in mitotic chromosomal ... [full text in table]

Curated tissue note. Highly expressed in hematopoietic tissues, fetal liver, spleen, thymus and adult thymus and bone marrow. Lower levels are found in heart, testis, kidney, colon and lung

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95239 | HPA portal (live; not the pinned values)

KIF6 | Unsafe / Protein

ENSG00000164627

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (cilia axoneme): Low (Enhanced); bronchus / ciliated cells (ciliary rootlets): High (Enhanced); bronchus / ciliated cells (tip of cilia): Low (Enhanced); caudate / glial cells: Medium (Enhanced); cerebral cortex / ... [full text in table]

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 24/99 (24.2%); Breast Invasive Carcinoma (validation): 10/50 (20.0%); Glioblastoma Multiforme (TCGA): 22/141 (15.6%)

Cancer IHC. glioma: 3/12 (25.0%); breast cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZMV9 | HPA portal (live; not the pinned values)

KIF9 | Unsafe / Protein

ENSG00000088727

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 495/495 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Rectum Adenocarcinoma (TCGA): 88/88 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); endometrial cancer: 12/12 (100.0%)

Biology. Essential for normal male fertility and for progressive motility of spermatozoa

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HAQ2 | HPA portal (live; not the pinned values)

KISS1 | Unsafe / RNA

ENSG00000170498

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 4.4 nTPM (limit 2); hypothalamus: 4 nTPM (limit 2)

Cancer RNA. Liver Hepatocellular Carcinoma (TCGA): 144/362 (39.8%); Liver Hepatocellular Carcinoma (validation): 67/231 (29.0%); Bladder Urothelial Carcinoma (TCGA): 49/169 (29.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Kisspeptins are ligands for the G protein-coupled receptor KISS1R/GPR54 (PubMed:11385580, PubMed:11457843, PubMed:11527393, PubMed:12879005, PubMed:15020672, PubMed:15596153). The hypothalamic KISS1/KISS1R signaling system plays a central role in the regulation of the hypothalamic-pituitary-gonadal reproductive axis by modulating the secretion of gonadotropin-releasing hormone (GnRH) from GnRH neurons (PubMed:15219839, PubMed:15598687, ... [full text in table]

Curated tissue note. High expression in placenta (PubMed:8944003, PubMed:11385580, PubMed:11457843, PubMed:12574233). Expression levels increased in both early placentas and molar pregnancies and are reduced in choriocarcinoma ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15726 | HPA portal (live; not the pinned values)

KLHL11 | Unsafe / Protein

ENSG00000178502

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 206/207 (99.5%); Colon Adenocarcinoma (validation): 480/486 (98.8%)

Cancer IHC. testis cancer: 7/8 (87.5%); renal cancer: 8/11 (72.7%)

Biology. Component of a cullin-RING-based BCR (BTB-CUL3-RBX1) E3 ubiquitin-protein ligase complex that mediates the ubiquitination of target proteins, leading most often to their proteasomal degradation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NVR0 | HPA portal (live; not the pinned values)

KLHL35 | Unsafe / RNA

ENSG00000149243

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.6 nTPM (limit 2); basal ganglia: 10.4 nTPM (limit 2); hippocampal formation: 3.7 nTPM (limit 2); hypothalamus: 5.8 nTPM (limit 2); midbrain: 2.6 nTPM (limit 2); spinal cord: 4.7 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 41/50 (82.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 177/283 (62.5%); Ovary Serous Cystadenocarcinoma (TCGA): 176/349 (50.4%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PF15 | HPA portal (live; not the pinned values)

KLHL7 | Unsafe / Protein

ENSG00000122550

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: High (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%)

Cancer IHC. colorectal cancer: 11/12 (91.7%); liver cancer: 10/12 (83.3%)

Biology. Substrate-specific adapter of a BCR (BTB-CUL3-RBX1) E3 ubiquitin ligase complex. The BCR(KLHL7) complex acts by mediating ubiquitination and subsequent degradation of substrate proteins. Probably mediates 'Lys-48'-linked ubiquitination

Curated tissue note. Widely expressed, with highest levels in adult and fetal heart, CNS and adult testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IXQ5 | HPA portal (live; not the pinned values)

KLK15 | Unsafe / High RNA

ENSG00000174562

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 109.2 nTPM; rectum/intestinal goblet cells; hpa_single_cell_tissue; protected; 0.2857 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Kidney Chromophobe (TCGA): 56/64 (87.5%); Prostate Adenocarcinoma (TCGA): 215/480 (44.8%); Rectum Adenocarcinoma (validation): 30/207 (14.5%)

Cancer IHC. prostate cancer: 3/10 (30.0%); skin cancer: 1/11 (9.1%)

Biology. Protease whose physiological substrate is not yet known

Curated tissue note. Highest expression in the thyroid gland. Also expressed in the prostate, salivary, and adrenal glands and in the colon testis and kidney

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H2R5 | HPA portal (live; not the pinned values)

KNL1 | Unsafe / Protein

ENSG00000137812

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); colon / glandular cells: Low (Enhanced); duodenum / glandular cells: Low (Enhanced); esophagus / squamous epithelial cells: Low (Enhanced); lymph node / non-germinal center cells: Low (Enhanced); rectum / glandular cells: Low (Enhanced); skin 1 ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 472/486 (97.1%); Glioblastoma Multiforme (validation): 56/58 (96.6%); Rectum Adenocarcinoma (validation): 191/207 (92.3%)

Cancer IHC. cervical cancer: 9/12 (75.0%); colorectal cancer: 4/11 (36.4%)

Biology. Acts as a component of the outer kinetochore KNL1 complex that serves as a docking point for spindle assembly checkpoint components and mediates microtubule-kinetochore interactions (PubMed:15502821, PubMed:17981135, PubMed:18045986, PubMed:19893618, PubMed:21199919, PubMed:22000412, PubMed:22331848, PubMed:27881301, PubMed:30100357). Kinetochores, consisting of a centromere-associated inner segment and a microtubule-contacting outer ... [full text in table]

Curated tissue note. Highly expressed in testis, where it is localized in germ cells, in particular in spermatocytes and in the pre-acrosome of round spermatids. Detected in the acrosome of ejaculated spermatozoa. Detected in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NG31 | HPA portal (live; not the pinned values)

KPNA2 | Unsafe / Protein

ENSG00000182481

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / ciliated cells (cell body): Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / Purkinje cells: Medium (Enhanced); cerebral cortex / glial cells: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Functions in nuclear protein import as an adapter protein for nuclear receptor KPNB1 (PubMed:28991411, PubMed:32130408, PubMed:7604027, PubMed:7754385). Binds specifically and directly to substrates containing either a simple or bipartite NLS motif (PubMed:28991411, PubMed:32130408, PubMed:7604027, PubMed:7754385). Docking of the importin/substrate complex to the nuclear pore complex (NPC) is mediated by KPNB1 through binding to ... [full text in table]

Curated tissue note. Expressed ubiquitously

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P52292 | HPA portal (live; not the pinned values)

KPNA5 | Unsafe / Protein

ENSG00000196911

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / cells in granular layer: Low (Enhanced); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. endometrial cancer: 2/12 (16.7%); colorectal cancer: 1/10 (10.0%)

Biology. Functions in nuclear protein import as an adapter protein for nuclear receptor KPNB1. Binds specifically and directly to substrates containing either a simple or bipartite NLS motif. Docking of the importin/substrate complex to the nuclear pore complex (NPC) is mediated by KPNB1 through binding to nucleoporin FxFG repeats and the complex is subsequently translocated through the pore by an energy requiring, Ran-dependent mechanism. At ... [full text in table]

Curated tissue note. Testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O15131 | HPA portal (live; not the pinned values)

KRBOX1 | Unsafe / RNA

ENSG00000240747

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.5 nTPM (limit 2); amygdala: 3.9 nTPM (limit 2); basal ganglia: 3.2 nTPM (limit 2); cerebellum: 2.2 nTPM (limit 2); cerebral cortex: 3.2 nTPM (limit 2); choroid plexus: 2.3 nTPM (limit 2); heart muscle: 3.6 nTPM (limit 2); hippocampal formation: 3.3 nTPM (limit 2); hypothalamus: 3.2 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 69/141 (48.9%); Thyroid Carcinoma (TCGA): 240/495 (48.5%); Ovary Serous Cystadenocarcinoma (TCGA): 159/349 (45.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt C9JBD0 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

KRIT1 | Unsafe / Protein

ENSG00000001631

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / enterocytes: Low (Supported); colon / goblet cells: Low (Supported); duodenum / enterocytes: Low (Supported); rectum / endothelial cells: Low (Supported)

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. colorectal cancer: 1/12 (8.3%); ovarian cancer: 1/12 (8.3%)

Biology. Component of the CCM signaling pathway which is a crucial regulator of heart and vessel formation and integrity (By similarity). Negative regulator of angiogenesis. Inhibits endothelial proliferation, apoptosis, migration, lumen formation and sprouting angiogenesis in primary endothelial cells. Promotes AKT phosphorylation in a NOTCH-dependent and independent manner, and inhibits ERK1/2 phosphorylation indirectly through activation of ... [full text in table]

Curated tissue note. Low levels in brain. Very weak expression found in heart and muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00522 | HPA portal (live; not the pinned values)

KRT25 | Unsafe / RNA

ENSG00000204897

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 108.8 nTPM (limit 10); skin: 108.8 nTPM (baseline 10)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 3/99 (3.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 4/176 (2.3%); Kidney Renal Clear Cell Carcinoma (validation): 1/100 (1.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Essential for the proper assembly of type I and type II keratin protein complexes and formation of keratin intermediate filaments in the inner root sheath (irs) (By similarity). Plays a role in the cytoskeleton organization (PubMed:26902920)

Curated tissue note. Strongly expressed in skin and scalp, and weak expression observed in thymus and tongue. In the hair follicle, expressed in Henle layer, Huxley layer and in the inner root sheath cuticle of the hair follicle. ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z3Z0 | HPA portal (live; not the pinned values)

KRT28 | Unsafe / RNA

ENSG00000173908

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 11.3 nTPM (limit 10); skin: 11.3 nTPM (baseline 10)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 5/176 (2.8%); Bladder Urothelial Carcinoma (TCGA): 2/169 (1.2%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 3/283 (1.1%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/9 (0.0%)

Biology. Essential for the proper assembly of types I and II keratin protein complexes and the formation of keratin intermediate filaments in the inner root sheath (irs)

Curated tissue note. Strongly expressed in skin and scalp, and weak expression observed in thymus. In the hair follicle, expressed in Henle layer, Huxley layer and in the irs cuticle. Expression extends from the bulb region up to ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z3Y7 | HPA portal (live; not the pinned values)

KRT33A | Unsafe / RNA

ENSG00000006059

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 46 nTPM (limit 10); skin: 46 nTPM (baseline 10)

Cancer RNA. Breast Invasive Carcinoma (validation): 1/50 (2.0%); Lung Squamous Cell Carcinoma (TCGA): 9/489 (1.8%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%)

Cancer IHC. testis cancer: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. Expressed in the hair follicles

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O76009 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

KRT33B | Unsafe / RNA

ENSG00000131738

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 80.2 nTPM (limit 10); skin: 80.2 nTPM (baseline 10)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Colon Adenocarcinoma (validation): 1/486 (0.2%)

Cancer IHC. testis cancer: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14525 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

KRT35 | Unsafe / RNA

ENSG00000197079

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. ; skin: 56.3 nTPM (baseline 10)

Cancer RNA. Breast Invasive Carcinoma (validation): 1/50 (2.0%); Rectum Adenocarcinoma (validation): 4/207 (1.9%); Colon Adenocarcinoma (validation): 9/486 (1.9%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/9 (0.0%)

Biology. Early expression in the hair follicle, mainly found in supramatricial cells and lowermost cortical cells of the hair bulb

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92764 | HPA portal (live; not the pinned values)

KRT72 | Unsafe / RNA

ENSG00000170486

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. skin: 10.2 nTPM (limit 10); skin: 10.2 nTPM (baseline 10); small intestine: 2.1 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 2/50 (4.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 3/176 (1.7%); Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Has a role in hair formation. Specific component of keratin intermediate filaments in the inner root sheath (IRS) of the hair follicle (Probable)

Curated tissue note. Highly expressed in hair follicles from scalp and eyebrow. Also expressed in palmoplantar epidermis. Not expressed in face skin despite the presence of fine hairs histologically. In hair, it is specifically ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14CN4 | HPA portal (live; not the pinned values)

KRT82 | Unsafe / RNA

ENSG00000161850

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. ; skin: 12.3 nTPM (baseline 10)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Structural component of intermediate filaments in the hair follicle. As a type II hair keratin, forms obligate heterodimers with type I hair keratins that assemble into keratin intermediate filaments. Expressed specifically in cuticle-forming cells of the hair shaft during the anagen phase, where it contributes to the formation and structural integrity of the hair cuticle

Curated tissue note. Expressed in the hair follicle, specifically in hair shaft cuticle during the anagen phase of the hair cycle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NSB4 | HPA portal (live; not the pinned values)

KRTAP10-1 | Unsafe / RNA

ENSG00000215455

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. skin: 10.1 nTPM (limit 10); skin: 10.1 nTPM (baseline 10)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. In the hair cortex, hair keratin intermediate filaments are embedded in an interfilamentous matrix, consisting of hair keratin-associated proteins (KRTAP), which are essential for the formation of a rigid and resistant hair shaft through their extensive disulfide bond cross-linking with abundant cysteine residues of hair keratins. The matrix proteins include the high-sulfur and high-glycine-tyrosine keratins

Curated tissue note. Restricted to a narrow region of the hair fiber cuticle, lying approximately 20 cell layers above the apex of the dermal papilla of the hair root; not detected in any other tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P60331 | HPA portal (live; not the pinned values)

KRTAP10-2 | Unsafe / RNA

ENSG00000205445

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 14.6 nTPM (limit 10); skin: 14.6 nTPM (baseline 10)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. In the hair cortex, hair keratin intermediate filaments are embedded in an interfilamentous matrix, consisting of hair keratin-associated proteins (KRTAP), which are essential for the formation of a rigid and resistant hair shaft through their extensive disulfide bond cross-linking with abundant cysteine residues of hair keratins. The matrix proteins include the high-sulfur and high-glycine-tyrosine keratins

Curated tissue note. Restricted to a narrow region of the hair fiber cuticle, lying approximately 20 cell layers above the apex of the dermal papilla of the hair root; not detected in any other tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P60368 | HPA portal (live; not the pinned values)

KRTAP10-7 | Unsafe / RNA

ENSG00000272804

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 13.8 nTPM (limit 10); skin: 13.8 nTPM (baseline 10)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. In the hair cortex, hair keratin intermediate filaments are embedded in an interfilamentous matrix, consisting of hair keratin-associated proteins (KRTAP), which are essential for the formation of a rigid and resistant hair shaft through their extensive disulfide bond cross-linking with abundant cysteine residues of hair keratins. The matrix proteins include the high-sulfur and high-glycine-tyrosine keratins

Curated tissue note. Restricted to a narrow region of the hair fiber cuticle, lying approximately 20 cell layers above the apex of the dermal papilla of the hair root; not detected in any other tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P60409 | HPA portal (live; not the pinned values)

KRTAP26-1 | Unsafe / RNA

ENSG00000197683

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 18.7 nTPM (limit 10); skin: 18.7 nTPM (baseline 10)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Prostate Adenocarcinoma (TCGA): 1/480 (0.2%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. In the hair cortex, hair keratin intermediate filaments are embedded in an interfilamentous matrix, consisting of hair keratin-associated proteins (KRTAP), which are essential for the formation of a rigid and resistant hair shaft through their extensive disulfide bond cross-linking with abundant cysteine residues of hair keratins. The matrix proteins include the high-sulfur and high-glycine-tyrosine keratins

Curated tissue note. Localized high up in the well differentiated portion of the hair follicle cuticle (about 10-15 cell layers above the apex of the dermal papilla)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PEX3 | HPA portal (live; not the pinned values)

KRTAP5-8 | Unsafe / RNA

ENSG00000241233

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 5.8 nTPM (limit 2); kidney: 5.8 nTPM (baseline 2)

Cancer RNA. Kidney Chromophobe (TCGA): 52/64 (81.2%); Bladder Urothelial Carcinoma (TCGA): 6/169 (3.6%); Kidney Renal Clear Cell Carcinoma (TCGA): 16/521 (3.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. In the hair cortex, hair keratin intermediate filaments are embedded in an interfilamentous matrix, consisting of hair keratin-associated protein (KRTAP), which are essential for the formation of a rigid and resistant hair shaft through their extensive disulfide bond cross-linking with abundant cysteine residues of hair keratins. The matrix proteins include the high-sulfur and high-glycine-tyrosine keratins

Curated tissue note. Restricted to hair root, not detected in any other tissues. Expressed in cuticle layers of differentiating hair follicles

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75690 | HPA portal (live; not the pinned values)

KRTAP8-1 | Unsafe / RNA

ENSG00000183640

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 59.4 nTPM (limit 10); skin: 59.4 nTPM (baseline 10)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 1/99 (1.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. In the hair cortex, hair keratin intermediate filaments are embedded in an interfilamentous matrix, consisting of hair keratin-associated proteins (KRTAP), which are essential for the formation of a rigid and resistant hair shaft through their extensive disulfide bond cross-linking with abundant cysteine residues of hair keratins. The matrix proteins include the high-sulfur and high-glycine-tyrosine keratins

Curated tissue note. Is essentially restricted to only one vertical half of the hair forming compartment and in beard hairs is absent from the central medulla

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IUC2 | HPA portal (live; not the pinned values)

KRTAP9-2 | Unsafe / RNA

ENSG00000239886

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 31.7 nTPM (limit 10); skin: 31.7 nTPM (baseline 10)

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 0/283 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. In the hair cortex, hair keratin intermediate filaments are embedded in an interfilamentous matrix, consisting of hair keratin-associated proteins (KRTAP), which are essential for the formation of a rigid and resistant hair shaft through their extensive disulfide bond cross-linking with abundant cysteine residues of hair keratins. The matrix proteins include the high-sulfur and high-glycine-tyrosine keratins

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BYQ4 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

L1TD1 | Unsafe / High RNA

ENSG00000240563

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 57.6 nTPM; colon/intestinal goblet cells; hpa_single_cell_tissue; protected; 57.22 TPM; blood/M2; dice_m2; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 125/133 (94.0%); Colon Adenocarcinoma (validation): 222/486 (45.7%); Colon Adenocarcinoma (TCGA): 96/254 (37.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. RNA-binding protein which plays a role in the regulation of pluripotency. Preferentially binds to 5'-GAGCGU-3' sequences. Binds to the 3'-UTR of the pluripotency regulator TFCP2L1, stabilizing transcripts and resulting in increased TFCP2L1 mRNA levels

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T7N2 | HPA portal (live; not the pinned values)

L3MBTL4 | Unsafe / Protein

ENSG00000154655

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebral cortex / glial cells: Low (Approved); colon / peripheral nerve/ganglion: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 46/58 (79.3%); Kidney Renal Clear Cell Carcinoma (validation): 70/100 (70.0%); Liver Hepatocellular Carcinoma (validation): 160/231 (69.3%)

Cancer IHC. lung cancer: 5/12 (41.7%); liver cancer: 3/10 (30.0%)

Biology. Putative Polycomb group (PcG) protein. PcG proteins maintain the transcriptionally repressive state of genes, probably via a modification of chromatin, rendering it heritably changed in its expressibility (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NA19 | HPA portal (live; not the pinned values)

LACTB | Unsafe / Protein

ENSG00000103642

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: Medium (Supported); bronchus / ciliated cells (cell body): Medium (Supported); caudate / glial cells: Low (Supported); caudate / neuronal cells: Medium (Supported); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%)

Cancer IHC. liver cancer: 11/11 (100.0%); melanoma: 11/12 (91.7%)

Biology. Mitochondrial serine protease that acts as a regulator of mitochondrial lipid metabolism (PubMed:28329758). Acts by decreasing protein levels of PISD, a mitochondrial enzyme that converts phosphatidylserine (PtdSer) to phosphatidylethanolamine (PtdEtn), thereby affecting mitochondrial lipid metabolism (PubMed:28329758). It is unclear whether it acts directly by mediating proteolysis of PISD or by mediating proteolysis of another lipid ... [full text in table]

Curated tissue note. Expressed predominantly in skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P83111 | HPA portal (live; not the pinned values)

LACTBL1 | Unsafe / RNA

ENSG00000215906

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 3.8 nTPM (limit 2); retina: 3.8 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A8MY62 | HPA portal (live; not the pinned values)

LALBA | Unsafe / High RNA

ENSG00000167531

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 1303 nTPM; breast/macrophages; hpa_single_cell_tissue; vital; 0.5688 TPM; blood/TH17; dice_th17; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 5/50 (10.0%); Breast Invasive Carcinoma (TCGA): 28/1022 (2.7%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Regulatory subunit of lactose synthase, changes the substrate specificity of galactosyltransferase in the mammary gland making glucose a good acceptor substrate for this enzyme. This enables LS to synthesize lactose, the major carbohydrate component of milk. In other tissues, galactosyltransferase transfers galactose onto the N-acetylglucosamine of the oligosaccharide chains in glycoproteins

Curated tissue note. Mammary gland specific. Secreted in milk

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P00709 | HPA portal (live; not the pinned values)

LCA5 | Unsafe / Protein

ENSG00000135338

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Supported); bone marrow / hematopoietic cells: Medium (Supported); bronchus / ciliated cells (tip of cilia): High (Supported); caudate / glial cells: Medium (Supported); cerebral cortex / glial cells: Medium (Supported); cerebral cortex / neuropil: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Thyroid Carcinoma (TCGA): 373/495 (75.4%)

Cancer IHC. prostate cancer: 11/11 (100.0%); breast cancer: 8/12 (66.7%)

Biology. Involved in intraflagellar protein (IFT) transport in photoreceptor cilia. Plays a role in the ciliary transport of photoreceptors outer segment proteins

Curated tissue note. Widely expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86VQ0 | HPA portal (live; not the pinned values)

LCA5L | Unsafe / Protein

ENSG00000157578

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); kidney / cells in glomeruli: Medium (Enhanced); kidney / distal tubules: Low (Enhanced); kidney / proximal tubules (microvilli): Medium (Enhanced); nasopharynx / ciliated cells (ciliary rootlets): Low (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 46/50 (92.0%); Glioblastoma Multiforme (validation): 11/58 (19.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 21/176 (11.9%)

Cancer IHC. glioma: 7/12 (58.3%); melanoma: 3/9 (33.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95447 | HPA portal (live; not the pinned values)

LCE2D | Unsafe / RNA

ENSG00000187223

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skeletal muscle: 2.1 nTPM (limit 2); skin: 147.2 nTPM (limit 10); skeletal muscle: 2.1 nTPM (baseline 2); skin: 147.2 nTPM (baseline 10)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 24/99 (24.2%); Head and Neck Squamous Cell Carcinoma (TCGA): 48/492 (9.8%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Precursors of the cornified envelope of the stratum corneum

Curated tissue note. Skin-specific. Expression was readily detected in adult trunk skin, adult arm skin, fetal skin, penal skin, vulva, esophagus and tongue. Not expressed in the cervix, rectum, lung, colon, or placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5TA82 | HPA portal (live; not the pinned values)

LCOR | Unsafe / Protein

ENSG00000196233

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. breast cancer: 11/11 (100.0%); colorectal cancer: 11/11 (100.0%)

Biology. May act as transcription activator that binds DNA elements with the sequence 5'-CCCTATCGATCGATCTCTACCT-3' (By similarity). Repressor of ligand-dependent transcription activation by target nuclear receptors. Repressor of ligand-dependent transcription activation by ESR1, ESR2, NR3C1, PGR, RARA, RARB, RARG, RXRA and VDR

Curated tissue note. Ubiquitous

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96JN0 | HPA portal (live; not the pinned values)

LCORL | Unsafe / Protein

ENSG00000178177

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 479/486 (98.6%)

Cancer IHC. liver cancer: 4/12 (33.3%); pancreatic cancer: 2/11 (18.2%)

Biology. May act as transcription activator that binds DNA elements with the sequence 5'-CCCTATCGATCGATCTCTACCT-3'. May play a role in spermatogenesis (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N3X6 | HPA portal (live; not the pinned values)

LELP1 | Unsafe / High RNA

ENSG00000203784

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 65.7 nTPM; testis/macrophages; hpa_single_cell_tissue; vital; 0 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Lung Squamous Cell Carcinoma (TCGA): 1/489 (0.2%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T871 | HPA portal (live; not the pinned values)

LEMD1 | Unsafe / RNA

ENSG00000186007

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 3.6 nTPM (limit 2); cerebellum: 3.6 nTPM (baseline 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 284/349 (81.4%); Ovary Serous Cystadenocarcinoma (validation): 65/81 (80.2%); Pancreatic Adenocarcinoma (validation): 61/80 (76.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Testis-specific. Isoform 6 is detected in 17 of 18 colon cancer tissues examined

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q68G75 | HPA portal (live; not the pinned values)

LGALS8 | Unsafe / Protein

ENSG00000116977

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%)

Cancer IHC. breast cancer: 11/11 (100.0%); colorectal cancer: 11/12 (91.7%)

Biology. Beta-galactoside-binding lectin that acts as a sensor of membrane damage caused by infection and restricts the proliferation of infecting pathogens by targeting them for autophagy (PubMed:22246324, PubMed:28077878). Detects membrane rupture by binding beta-galactoside ligands located on the lumenal side of the endosome membrane; these ligands becoming exposed to the cytoplasm following rupture (PubMed:22246324, PubMed:28077878). ... [full text in table]

Curated tissue note. Ubiquitous. Selective expression by prostate carcinomas versus normal prostate and benign prostatic hypertrophy

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00214 | HPA portal (live; not the pinned values)

LHX9 | Unsafe / RNA

ENSG00000143355

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 3.6 nTPM (limit 2); retina: 3.6 nTPM (baseline 2)

Cancer RNA. Kidney Chromophobe (TCGA): 37/64 (57.8%); Glioblastoma Multiforme (validation): 22/58 (37.9%); Glioblastoma Multiforme (TCGA): 21/141 (14.9%)

Cancer IHC. cervical cancer: 2/11 (18.2%); colorectal cancer: 2/12 (16.7%)

Biology. Involved in gonadal development

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NQ69 | HPA portal (live; not the pinned values)

LIAT1 | Unsafe / Protein

ENSG00000187624

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebral cortex / glial cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); cerebral cortex / neuropil: Low (Approved); colon / glandular cells: Low (Approved); colon / ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 45/50 (90.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 243/282 (86.2%); Glioblastoma Multiforme (validation): 37/58 (63.8%)

Cancer IHC. breast cancer: 9/12 (75.0%); endometrial cancer: 8/12 (66.7%)

Biology. Participates in nucleolar liquid-liquid phase separation (LLPS) through its N-terminal intrinsically disordered region (IDR). May be involved in ATE1-mediated N-terminal arginylation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZQX7 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

LIG4 | Unsafe / Protein

ENSG00000174405

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Adenocarcinoma (validation): 104/105 (99.0%); Rectum Adenocarcinoma (validation): 199/207 (96.1%)

Cancer IHC. prostate cancer: 10/12 (83.3%); glioma: 9/12 (75.0%)

Biology. DNA ligase involved in DNA non-homologous end joining (NHEJ); required for double-strand break (DSB) repair and V(D)J recombination (PubMed:12517771, PubMed:17290226, PubMed:23523427, PubMed:29980672, PubMed:33586762, PubMed:8798671, PubMed:9242410, PubMed:9809069). Catalyzes the NHEJ ligation step of the broken DNA during DSB repair by resealing the DNA breaks after the gap filling is completed (PubMed:12517771, PubMed:17290226, ... [full text in table]

Curated tissue note. Testis, thymus, prostate and heart

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P49917 | HPA portal (live; not the pinned values)

LIN28A | Unsafe / Protein

ENSG00000131914

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced); colon / glandular cells: Low (Enhanced); duodenum / glandular cells: Low (Enhanced); heart muscle / cardiomyocytes: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); lung / alveolar cells: Low (Enhanced); rectum / glandular cells: ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 126/133 (94.7%); Breast Invasive Carcinoma (validation): 8/50 (16.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 8/176 (4.5%)

Cancer IHC. testis cancer: 11/12 (91.7%); skin cancer: 3/10 (30.0%)

Biology. RNA-binding protein that inhibits processing of pre-let-7 miRNAs and regulates translation of mRNAs that control developmental timing, pluripotency and metabolism (PubMed:21247876). Seems to recognize a common structural G-quartet (G4) feature in its miRNA and mRNA targets (Probable). 'Translational enhancer' that drives specific mRNAs to polysomes and increases the efficiency of protein synthesis. Its association with the ... [full text in table]

Curated tissue note. Expressed in embryonic stem cells, placenta and testis. Tends to be up-regulated in HER2-overexpressing breast tumors

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H9Z2 | HPA portal (live; not the pinned values)

LIN28B | Unsafe / High RNA

ENSG00000187772

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 65.8 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 0.9 TPM; blood/Exhausted memory B-cell; hpa_immune_monaco; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 113/133 (85.0%); Glioblastoma Multiforme (validation): 7/58 (12.1%); Liver Hepatocellular Carcinoma (validation): 27/231 (11.7%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Suppressor of microRNA (miRNA) biogenesis, including that of let-7 and possibly of miR107, miR-143 and miR-200c. Binds primary let-7 transcripts (pri-let-7), including pri-let-7g and pri-let-7a-1, and sequester them in the nucleolus, away from the microprocessor complex, hence preventing their processing into mature miRNA (PubMed:22118463). Does not act on pri-miR21 (PubMed:22118463). The repression of let-7 expression is required for ... [full text in table]

Curated tissue note. Expressed at high levels in the placenta and, at mucher lower, in testis and fetal liver (PubMed:16971064). Isoform 1 is only detected in placenta and in moderately and poorly differentiated hepatocellular ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZN17 | HPA portal (live; not the pinned values)

LIN7A | Unsafe / Protein

ENSG00000111052

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Low (Approved); colon / peripheral nerve/ganglion: Low (Approved); duodenum / glandular cells: Low (Approved); hippocampus / neuronal cells: Low (Approved); kidney / cells in tubules: Medium (Approved); liver / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Kidney Chromophobe (TCGA): 61/64 (95.3%); Liver Hepatocellular Carcinoma (validation): 203/231 (87.9%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Plays a role in establishing and maintaining the asymmetric distribution of channels and receptors at the plasma membrane of polarized cells. Forms membrane-associated multiprotein complexes that may regulate delivery and recycling of proteins to the correct membrane domains. The tripartite complex composed of LIN7 (LIN7A, LIN7B or LIN7C), CASK and APBA1 associates with the motor protein KIF17 to transport vesicles containing ... [full text in table]

Curated tissue note. Expressed in brain, testis, kidney, placenta and liver

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14910 | HPA portal (live; not the pinned values)

LIPT1 | Unsafe / Protein

ENSG00000144182

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bronchus / ciliated cells (ciliary rootlets): Medium (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: Low (Approved); colon / peripheral nerve/ganglion: Low (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Squamous Cell Carcinoma (validation): 67/68 (98.5%)

Cancer IHC. endometrial cancer: 1/11 (9.1%); melanoma: 1/11 (9.1%)

Biology. Lipoyl amidotransferase that catalyzes the transfer of lipoyl moieties from lipoyl-protein H of the glycine cleavage system (lipoyl-GCSH) to E2 subunits of the pyruvate dehydrogenase complex (PDCE2) (PubMed:29987032). Unable to catalyze the transfer of octanoyl from octanoyl-GCSH to PDCE2 (PubMed:29987032). In vitro, it is also able to catalyze the transfer of the lipoyl group from lipoyl-AMP to the specific lysine residue of lipoyl ... [full text in table]

Curated tissue note. Highly expressed in skeletal muscle and heart, moderately in kidney and pancreas, and detected at lower levels in liver, brain, placenta and lung

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y234 | HPA portal (live; not the pinned values)

LIPT2 | Unsafe / Protein

ENSG00000175536

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: High (Approved); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Ovary Serous Cystadenocarcinoma (TCGA): 242/349 (69.3%); Kidney Renal Clear Cell Carcinoma (validation): 55/100 (55.0%)

Cancer IHC. breast cancer: 11/12 (91.7%); colorectal cancer: 11/12 (91.7%)

Biology. Catalyzes the transfer of endogenously produced octanoic acid from octanoyl-acyl-carrier-protein (octanoyl-ACP) onto the lipoyl domains of lipoate-dependent enzymes such as the protein H of the glycine cleavage system (GCSH) (PubMed:28757203). Lipoyl-ACP can also act as a substrate although octanoyl-ACP is likely to be the physiological substrate (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NK58 | HPA portal (live; not the pinned values)

LMX1B | Unsafe / RNA

ENSG00000136944

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 3.5 nTPM (limit 2); midbrain: 3.6 nTPM (limit 2); salivary gland: 18 nTPM (limit 6); kidney: 3.5 nTPM (baseline 2); midbrain: 3.6 nTPM (baseline 2); salivary gland: 18 nTPM (baseline 6)

Cancer RNA. Breast Invasive Carcinoma (validation): 49/50 (98.0%); Breast Invasive Carcinoma (TCGA): 822/1022 (80.4%); Skin Cuteneous Melanoma (TCGA): 6/99 (6.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcription factor involved in the regulation of podocyte-expressed genes (PubMed:24042019, PubMed:28059119). Essential for the specification of dorsal limb fate at both the zeugopodal and autopodal levels

Curated tissue note. Expressed in most tissues. Highest levels in testis, thyroid, duodenum, skeletal muscle, and pancreatic islets

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O60663 | HPA portal (live; not the pinned values)

LOXHD1 | Unsafe / RNA

ENSG00000167210

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.8 nTPM (limit 2); kidney: 6.7 nTPM (limit 2); bone marrow: 2.8 nTPM (baseline 2); kidney: 6.7 nTPM (baseline 2)

Cancer RNA. Kidney Chromophobe (TCGA): 7/64 (10.9%); Testicular Germ Cell Tumor (TCGA): 11/133 (8.3%); Breast Invasive Carcinoma (validation): 2/50 (4.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Involved in hearing. Required for normal function of hair cells in the inner ear (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IVV2 | HPA portal (live; not the pinned values)

LRAT | Unsafe / RNA

ENSG00000121207

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. liver: 2.1 nTPM (limit 2); liver: 2.1 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 20/58 (34.5%); Pancreatic Adenocarcinoma (validation): 13/80 (16.2%); Breast Invasive Carcinoma (validation): 6/50 (12.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transfers the acyl group from the sn-1 position of phosphatidylcholine to all-trans retinol, producing all-trans retinyl esters (PubMed:9920938). Retinyl esters are storage forms of vitamin A (Probable). LRAT plays a critical role in vision (Probable). It provides the all-trans retinyl ester substrates for the isomerohydrolase which processes the esters into 11-cis-retinol in the retinal pigment epithelium; due to a membrane-associated ... [full text in table]

Curated tissue note. Hepatic stellate cells and endothelial cells (at protein level). Found at high levels in testis and liver, followed by retinal pigment epithelium, small intestine, prostate, pancreas and colon. Low expression ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95237 | HPA portal (live; not the pinned values)

LRFN4 | Unsafe / Protein

ENSG00000173621

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: High (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in molecular layer: Medium (Enhanced); cerebellum / Purkinje cells: High (Enhanced); cerebral cortex / glial cells: Medium (Enhanced); cerebral cortex / neuronal cells: High (Enhanced); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (TCGA): 88/88 (100.0%); Ovary Serous Cystadenocarcinoma (validation): 81/81 (100.0%); Breast Invasive Carcinoma (validation): 50/50 (100.0%)

Cancer IHC. glioma: 8/11 (72.7%); liver cancer: 2/12 (16.7%)

Biology. Promotes neurite outgrowth in hippocampal neurons. May play a role in redistributing DLG4 to the cell periphery (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PJG9 | HPA portal (live; not the pinned values)

LRGUK | Unsafe / RNA

ENSG00000155530

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3.2 nTPM (limit 2); retina: 2.5 nTPM (limit 2); choroid plexus: 3.2 nTPM (baseline 2); retina: 2.5 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 51/58 (87.9%); Breast Invasive Carcinoma (validation): 17/50 (34.0%); Skin Cuteneous Melanoma (TCGA): 13/99 (13.1%)

Cancer IHC. breast cancer: 8/12 (66.7%); prostate cancer: 7/12 (58.3%)

Biology. Involved in multiple aspects of sperm assembly including acrosome attachment, shaping of the sperm head and in the early aspects of axoneme development. Not essential for primary cilium biogenesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96M69 | HPA portal (live; not the pinned values)

LRIT1 | Unsafe / RNA

ENSG00000148602

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 24.9 nTPM (limit 2); retina: 24.9 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Photoreceptor synaptic protein essential for normal vision (By similarity). Involved in synapse formation in cone photoreceptor cells (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P2V4 | HPA portal (live; not the pinned values)

LRP1B | Unsafe / Protein

ENSG00000168702

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebral cortex / neuronal cells: Medium (Approved); cerebral cortex / neuropil: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 71/141 (50.4%); Breast Invasive Carcinoma (validation): 12/50 (24.0%)

Cancer IHC. liver cancer: 8/11 (72.7%); colorectal cancer: 3/12 (25.0%)

Biology. Potential cell surface proteins that bind and internalize ligands in the process of receptor-mediated endocytosis

Curated tissue note. Expressed in thyroid gland and in salivary gland, as well as in adult and fetal brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NZR2 | HPA portal (live; not the pinned values)

LRP2BP | Unsafe / Protein

ENSG00000109771

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / neuronal cells: Medium (Approved); colon / endothelial cells: Low (Approved); colon / peripheral nerve/ganglion: Low (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 36/58 (62.1%); Breast Invasive Carcinoma (validation): 17/50 (34.0%); Glioblastoma Multiforme (TCGA): 27/141 (19.1%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. May act as an adapter that regulates LRP2 function

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P2M1 | HPA portal (live; not the pinned values)

LRRC18 | Unsafe / Protein

ENSG00000165383

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): Low (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); nasopharynx / basal cells: Medium (Enhanced); nasopharynx / ciliated cells (tip of cilia): Medium (Enhanced)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 10/176 (5.7%); Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%); Lung Adenocarcinoma (TCGA): 8/497 (1.6%)

Cancer IHC. urothelial cancer: 1/11 (9.1%); breast cancer: 0/12 (0.0%)

Biology. May be involved in the regulation of spermatogenesis and sperm maturation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N456 | HPA portal (live; not the pinned values)

LRRC27 | Unsafe / Protein

ENSG00000148814

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Enhanced); cerebellum / Purkinje cells: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); hippocampus / neuronal cells: Low (Enhanced); kidney / distal tubules: Medium (Enhanced); kidney / proximal tubules (cell body): High (Enhanced); parathyroid ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 47/50 (94.0%); Kidney Renal Clear Cell Carcinoma (validation): 92/100 (92.0%); Thyroid Carcinoma (TCGA): 444/495 (89.7%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9C0I9 | HPA portal (live; not the pinned values)

LRRC28 | Unsafe / Protein

ENSG00000168904

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: High (Approved); cerebellum / ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86X40 | HPA portal (live; not the pinned values)

LRRC34 | Unsafe / Protein

ENSG00000171757

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / ciliated cells (cell body): Medium (Approved); bronchus / ciliated cells (cilia axoneme): Medium (Approved); bronchus / ciliated cells (tip of cilia): High (Approved); cerebellum / cells ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 47/58 (81.0%); Breast Invasive Carcinoma (validation): 28/50 (56.0%); Ovary Serous Cystadenocarcinoma (validation): 6/81 (7.4%)

Cancer IHC. pancreatic cancer: 2/12 (16.7%); colorectal cancer: 1/11 (9.1%)

Biology. Highly expressed in stem cells where it may be involved in regulation of pluripotency. In embryonic stem cells (ESCs), important for normal expression of the pluripotency regulators POU5F1/OCT4 and KLF4. Also important for expression of the ectodermal marker gene NES and the endodermal marker gene GATA4. Promotes stem cell proliferation in vitro

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZ02 | HPA portal (live; not the pinned values)

LRRC36 | Unsafe / High RNA

ENSG00000159708

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 105.6 nTPM; lung/alveolar cells type 2; hpa_single_cell_tissue; vital; 19.7 TPM; blood/plasmacytoid DC; hpa_immune_monaco; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 8/50 (16.0%); Glioblastoma Multiforme (validation): 6/58 (10.3%); Pancreatic Adenocarcinoma (validation): 4/80 (5.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q1X8D7 | HPA portal (live; not the pinned values)

LRRC37A2 | Unsafe / Protein

ENSG00000238083

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. small intestine / enterocytes - Gradient: Ascending (Supported)

Cancer RNA. Breast Invasive Carcinoma (validation): 48/50 (96.0%); Pancreatic Adenocarcinoma (validation): 69/80 (86.2%); Ovary Serous Cystadenocarcinoma (validation): 67/81 (82.7%)

Cancer IHC. liver cancer: 3/10 (30.0%); endometrial cancer: 1/11 (9.1%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NM11 | HPA portal (live; not the pinned values)

LRRC3B | Unsafe / Protein

ENSG00000179796

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / glial cells: Medium (Approved); cerebral cortex / neuronal cells: Medium (Approved); cerebral cortex / neuropil: Medium (Approved); skeletal muscle / myocytes: Medium (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 42/58 (72.4%); Glioblastoma Multiforme (TCGA): 77/141 (54.6%); Ovary Serous Cystadenocarcinoma (TCGA): 50/349 (14.3%)

Cancer IHC. breast cancer: 11/11 (100.0%); endometrial cancer: 11/11 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96PB8 | HPA portal (live; not the pinned values)

LRRC52 | Unsafe / RNA

ENSG00000162763

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3 nTPM (limit 2); choroid plexus: 3 nTPM (baseline 2)

Cancer RNA. Kidney Chromophobe (TCGA): 4/64 (6.2%); Lung Adenocarcinoma (TCGA): 7/497 (1.4%); Thyroid Carcinoma (TCGA): 5/495 (1.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Auxiliary protein of the large-conductance, voltage and calcium-activated potassium channel (BK alpha). Modulates gating properties by producing a marked shift in the BK channel's voltage dependence of activation in the hyperpolarizing direction, and in the absence of calcium. KCNU1 channel auxiliary protein. Modulates KCNU1 gating properties

Curated tissue note. Mainly expressed in testis and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N7C0 | HPA portal (live; not the pinned values)

LRRC63 | Unsafe / RNA

ENSG00000173988

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.3 nTPM (limit 2); spinal cord: 4 nTPM (limit 2); retina: 2.3 nTPM (baseline 2); spinal cord: 4 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 7/58 (12.1%); Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%); Skin Cuteneous Melanoma (TCGA): 2/99 (2.0%)

Cancer IHC. testis cancer: 11/12 (91.7%); melanoma: 5/12 (41.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q05C16 | HPA portal (live; not the pinned values)

LRRC71 | Unsafe / RNA

ENSG00000160838

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 11.1 nTPM (limit 2); lung: 2.3 nTPM (limit 2); choroid plexus: 11.1 nTPM (baseline 2); lung: 2.3 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 23/176 (13.1%); Glioblastoma Multiforme (validation): 3/58 (5.2%); Lung Adenocarcinoma (TCGA): 15/497 (3.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N4P6 | HPA portal (live; not the pinned values)

LRRC73 | Unsafe / Protein

ENSG00000204052

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): High (Enhanced); caudate / glial cells: Low (Enhanced); cerebellum / Purkinje cells - cytoplasm/membrane: Low (Enhanced); cerebellum / Purkinje cells - dendrites: Low (Enhanced); cerebellum / synaptic glomeruli - core: High (Enhanced); cerebral cortex / ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 42/176 (23.9%); Kidney Renal Papillary Cell Carcinoma (TCGA): 56/282 (19.9%); Ovary Serous Cystadenocarcinoma (TCGA): 51/349 (14.6%)

Cancer IHC. cervical cancer: 2/10 (20.0%); prostate cancer: 2/11 (18.2%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JTD7 | HPA portal (live; not the pinned values)

LRRC8D | Unsafe / RNA

ENSG00000171492

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 26 nTPM (limit 2); amygdala: 15 nTPM (limit 2); basal ganglia: 17.1 nTPM (limit 2); bone marrow: 14 nTPM (limit 2); cerebellum: 8.2 nTPM (limit 2); cerebral cortex: 20.7 nTPM (limit 2); choroid plexus: 7.7 nTPM (limit 2); colon: 17.3 nTPM (limit 6); duodenum: 18.5 nTPM (limit 2); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Non-essential component of the volume-regulated anion channel (VRAC, also named VSOAC channel), an anion channel required to maintain a constant cell volume in response to extracellular or intracellular osmotic changes (PubMed:24790029, PubMed:26530471, PubMed:26824658, PubMed:28193731, PubMed:32415200). The VRAC channel conducts iodide better than chloride and can also conduct organic osmolytes like taurine (PubMed:24790029, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7L1W4 | HPA portal (live; not the pinned values)

LRRC9 | Unsafe / RNA

ENSG00000131951

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.6 nTPM (limit 2); choroid plexus: 2.6 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 8/58 (13.8%); Prostate Adenocarcinoma (TCGA): 32/480 (6.7%); Kidney Chromophobe (TCGA): 4/64 (6.2%)

Cancer IHC. lung cancer: 2/12 (16.7%); prostate cancer: 1/10 (10.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZRR7 | HPA portal (live; not the pinned values)

LRRIQ1 | Unsafe / RNA

ENSG00000133640

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 13.6 nTPM (limit 2); hypothalamus: 2.2 nTPM (limit 2); lung: 2.5 nTPM (limit 2); retina: 8.5 nTPM (limit 2); choroid plexus: 13.6 nTPM (baseline 2); hypothalamus: 2.2 nTPM (baseline 2); lung: 2.5 nTPM (baseline 2); retina: 8.5 nTPM (baseline 2)

Cancer RNA. Kidney Renal Papillary Cell Carcinoma (TCGA): 66/282 (23.4%); Breast Invasive Carcinoma (validation): 8/50 (16.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 25/176 (14.2%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96JM4 | HPA portal (live; not the pinned values)

LRRIQ3 | Unsafe / Protein

ENSG00000162620

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): High (Approved); kidney / cells in tubules: Low (Approved); nasopharynx / ciliated cells (ciliary rootlets): High (Approved); parathyroid gland / glandular cells: Low (Approved)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 4/99 (4.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 11/282 (3.9%); Bladder Urothelial Carcinoma (TCGA): 6/169 (3.6%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6PVS8 | HPA portal (live; not the pinned values)

LRWD1 | Unsafe / Protein

ENSG00000161036

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. lung / macrophages: Medium (Enhanced)

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Ovary Serous Cystadenocarcinoma (TCGA): 348/349 (99.7%); Colon Adenocarcinoma (TCGA): 253/254 (99.6%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Required for G1/S transition. Recruits and stabilizes the origin recognition complex (ORC) onto chromatin during G1 to establish pre-replication complex (preRC) and to heterochromatic sites in post-replicated cells. Binds a combination of DNA and histone methylation repressive marks on heterochromatin. Binds histone H3 and H4 trimethylation marks H3K9me3, H3K27me3 and H4K20me3 in a cooperative manner with DNA methylation. Required for ... [full text in table]

Curated tissue note. Testis-specific. Drastically down-regulated in testis from patients with Sertoli cell-only syndrome (SCOS)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UFC0 | HPA portal (live; not the pinned values)

LSM12 | Unsafe / Protein

ENSG00000161654

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular layer: ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); testis cancer: 12/12 (100.0%)

Biology. Nicotinic acid adenine dinucleotide phosphate (NAADP) binding protein (PubMed:34362892). Confers NAADP sensitivity to the two pore channel complex (TPCs) by acting as TPC accessory protein necessary for NAADP-evoked Ca(2+) release (PubMed:34362892)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3MHD2 | HPA portal (live; not the pinned values)

LY6K | Unsafe / RNA

ENSG00000160886

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. 106.2 nTPM; tongue/suprabasal keratinocytes; hpa_single_cell_tissue; protected; 3.4 TPM; blood/neutrophil; hpa_immune_cell_samples; vital

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 475/492 (96.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 266/283 (94.0%); Lung Squamous Cell Carcinoma (TCGA): 378/489 (77.3%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Required for sperm migration into the oviduct and male fertility by controlling binding of sperm to zona pellucida (By similarity). May play a role in cell growth (PubMed:18089789)

Curated tissue note. Specifically expressed in testis (at protein level)

Therapeutic evidence. Trial registration only.

UniProt Q17RY6 | Trial/source record | HPA portal (live; not the pinned values)

M1AP | Unsafe / RNA

ENSG00000159374

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.4 nTPM (limit 2); bone marrow: 2.4 nTPM (baseline 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 150/349 (43.0%); Testicular Germ Cell Tumor (TCGA): 49/133 (36.8%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 78/283 (27.6%)

Cancer IHC. prostate cancer: 12/12 (100.0%); lymphoma: 11/12 (91.7%)

Biology. Required for meiosis I progression during spermatogenesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TC57 | HPA portal (live; not the pinned values)

MAEL | Unsafe / RNA

ENSG00000143194

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. heart muscle: 5.4 nTPM (limit 4); retina: 4.2 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 61/133 (45.9%); Glioblastoma Multiforme (validation): 24/58 (41.4%); Glioblastoma Multiforme (TCGA): 51/141 (36.2%)

Cancer IHC. prostate cancer: 1/10 (10.0%); breast cancer: 1/11 (9.1%)

Biology. Plays a central role during spermatogenesis by repressing transposable elements and preventing their mobilization, which is essential for the germline integrity. Acts via the piRNA metabolic process, which mediates the repression of transposable elements during meiosis by forming complexes composed of piRNAs and Piwi proteins and governs the methylation and subsequent repression of transposons. Its association with piP-bodies suggests ... [full text in table]

Curated tissue note. Testis-specific. Expressed in various cancer cell lines, probably due to demethylation of its promoter

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96JY0 | HPA portal (live; not the pinned values)

MAF | Unsafe / Protein

ENSG00000178573

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: Low (Approved); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 105/105 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); renal cancer: 11/11 (100.0%)

Biology. Acts as a transcriptional activator or repressor. Involved in embryonic lens fiber cell development. Recruits the transcriptional coactivators CREBBP and/or EP300 to crystallin promoters leading to up-regulation of crystallin gene during lens fiber cell differentiation. Activates the expression of IL4 in T helper 2 (Th2) cells. Increases T-cell susceptibility to apoptosis by interacting with MYB and decreasing BCL2 expression. Together ... [full text in table]

Curated tissue note. Expressed in endothelial cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75444 | HPA portal (live; not the pinned values)

MAGEA12 | Unsafe / Safety

ENSG00000213401

Decision. A separately reviewed safety concern overrides expression nomination.

Normal RNA / IHC. 1.9 nTPM; placenta/mixed immune cells; hpa_single_cell_tissue; vital; 0 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 46/99 (46.5%); Stomach Adenocarcinoma (TCGA): 88/346 (25.4%); Lung Squamous Cell Carcinoma (TCGA): 110/489 (22.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Not known, though may play a role tumor transformation or progression. In vitro promotes cell viability in melanoma cell lines

Curated tissue note. Expressed in many tumors of several types, such as melanoma, head and neck squamous cell carcinoma, lung carcinoma and breast carcinoma, but not in normal tissues except for testes

Therapeutic evidence. Clinical safety signal.

UniProt P43365 | Reviewed source 1 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

MAGEA3 | Unsafe / High RNA

ENSG00000221867

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 53.1 nTPM; colon/undifferentiated cells; hpa_single_cell_tissue; protected; 0.1664 TPM; blood/CD8_STIM; dice_cd8_stim; vital

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 55/99 (55.6%); Lung Squamous Cell Carcinoma (TCGA): 223/489 (45.6%); Bladder Urothelial Carcinoma (TCGA): 68/169 (40.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Activator of ubiquitin ligase activity of RING-type zinc finger-containing E3 ubiquitin-protein ligases that acts as a repressor of autophagy (PubMed:20864041, PubMed:31267705). May enhance ubiquitin ligase activity of TRIM28 and stimulate p53/TP53 ubiquitination by TRIM28. Proposed to act through recruitment and/or stabilization of the Ubl-conjugating enzyme (E2) at the E3:substrate complex (PubMed:17942928, PubMed:20864041). May play ... [full text in table]

Curated tissue note. Expressed in many tumors of several types, such as melanoma, head and neck squamous cell carcinoma, lung carcinoma and breast carcinoma, but not in normal tissues except for testes and placenta. Never ... [full text in table]

Reconsideration. Resolve the pinned high normal-cell signal and exact locus/protein identity before reconsideration.

Therapeutic evidence. Clinical safety signal | Negative randomized trial.

UniProt P43357 | Reviewed source 1 | Reviewed source 2 | Reviewed source 3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

MAGEB2 | Unsafe / High RNA

ENSG00000099399

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 118.4 nTPM; colon/undifferentiated cells; hpa_single_cell_tissue; protected; 2.154 TPM; blood/NK; dice_nk; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 98/133 (73.7%); Lung Squamous Cell Carcinoma (TCGA): 105/489 (21.5%); Skin Cuteneous Melanoma (TCGA): 14/99 (14.1%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May enhance ubiquitin ligase activity of RING-type zinc finger-containing E3 ubiquitin-protein ligases. Proposed to act through recruitment and/or stabilization of the Ubl-conjugating enzyme (E2) at the E3:substrate complex

Curated tissue note. Expressed in testis and placenta, and in a significant fraction of tumors of various histologic types

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O15479 | HPA portal (live; not the pinned values)

MAGEB4 | Unsafe / Protein

ENSG00000120289

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); duodenum / glandular cells: Low (Enhanced); lymph node / germinal center cells: Low (Enhanced); nasopharynx / respiratory epithelial cells: Low (Enhanced); rectum / glandular cells: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 1/50 (2.0%); Breast Invasive Carcinoma (TCGA): 11/1022 (1.1%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O15481 | HPA portal (live; not the pinned values)

MAK | Unsafe / RNA

ENSG00000111837

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 6.9 nTPM (limit 2); retina: 115.3 nTPM (limit 2); choroid plexus: 6.9 nTPM (baseline 2); retina: 115.3 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 10/50 (20.0%); Glioblastoma Multiforme (validation): 10/58 (17.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 16/176 (9.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Essential for the regulation of ciliary length and required for the long-term survival of photoreceptors (By similarity). Phosphorylates FZR1 in a cell cycle-dependent manner. Plays a role in the transcriptional coactivation of AR. Could play an important function in spermatogenesis. May play a role in chromosomal stability in prostate cancer cells

Curated tissue note. Expressed in prostate cancer cell lines at generally higher levels than in normal prostate epithelial cell lines. Isoform 1 is expressed in kidney, testis, lung, trachea, and retina. Isoform 2 is ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P20794 | HPA portal (live; not the pinned values)

MAP10 | Unsafe / Protein

ENSG00000212916

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 53/58 (91.4%); Lung Adenocarcinoma (validation): 24/105 (22.9%); Pancreatic Adenocarcinoma (validation): 9/80 (11.2%)

Cancer IHC. pancreatic cancer: 12/12 (100.0%); prostate cancer: 6/7 (85.7%)

Biology. Microtubule-associated protein (MAP) that plays a role in the regulation of cell division; promotes microtubule stability and participates in the organization of the spindle midzone and normal progress of cytokinesis

Curated tissue note. Expressed in different cell lines (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P2G4 | HPA portal (live; not the pinned values)

MAP6 | Unsafe / Protein

ENSG00000171533

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (cilia axoneme): Low (Enhanced); bronchus / ciliated cells (tip of cilia): Low (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: High (Enhanced); cerebellum / molecular layer cells - ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 139/141 (98.6%); Prostate Adenocarcinoma (TCGA): 444/480 (92.5%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Involved in microtubule stabilization in many cell types, including neuronal cells (By similarity). Specifically has microtubule cold stabilizing activity (By similarity). Involved in dendrite morphogenesis and maintenance by regulating lysosomal trafficking via its interaction with TMEM106B (PubMed:24357581). Regulates KIF5A-mediated axonal cargo transport (By similarity). Regulates axonal growth during neuron polarization (By ... [full text in table]

Curated tissue note. Expressed in brain (at protein level). Expressed in spinal cord. Isoform 2 expression is up-regulated in the prefrontal cortex (Brodmann's area 46) of patients with schizophrenia (postmortem brain study)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96JE9 | HPA portal (live; not the pinned values)

MAP7D2 | Unsafe / Protein

ENSG00000184368

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / Bergmann glia - cytoplasm/membrane: High (Enhanced); cerebellum / GLUC cells - cytoplasm/membrane: High (Enhanced); cerebellum / granular cells - cytoplasm/membrane: Medium (Enhanced); ... [full text in table]

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 70/100 (70.0%); Testicular Germ Cell Tumor (TCGA): 81/133 (60.9%); Kidney Renal Clear Cell Carcinoma (TCGA): 264/521 (50.7%)

Cancer IHC. colorectal cancer: 3/8 (37.5%); stomach cancer: 2/11 (18.2%)

Biology. Microtubule-stabilizing protein that plays a role in the control of cell motility and neurite outgrowth via direct binding to the microtubule (By similarity). Acts as a critical cofactor for kinesin transport. In the proximal axon, regulates kinesin-1 family members, KIF5A, KIF5B and KIF5C recruitment to microtubules and contributes to kinesin-1-mediated transport in the axons (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96T17 | HPA portal (live; not the pinned values)

MARCHF10 | Unsafe / RNA

ENSG00000173838

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 4.1 nTPM (limit 2); choroid plexus: 17.8 nTPM (limit 2); hippocampal formation: 2.8 nTPM (limit 2); hypothalamus: 2.9 nTPM (limit 2); pituitary gland: 12.3 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (TCGA): 28/141 (19.9%); Uterine Corpus Endometrial Carcinoma (TCGA): 29/176 (16.5%); Kidney Chromophobe (TCGA): 10/64 (15.6%)

Cancer IHC. glioma: 5/12 (41.7%); breast cancer: 1/12 (8.3%)

Biology. E3 ubiquitin-protein ligase (Probable). E3 ubiquitin ligases accept ubiquitin from an E2 ubiquitin-conjugating enzyme in the form of a thioester and then directly transfer the ubiquitin to targeted substrates

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NA82 | HPA portal (live; not the pinned values)

MARCHF11 | Unsafe / RNA

ENSG00000183654

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.3 nTPM (limit 2); basal ganglia: 4.3 nTPM (limit 2); cerebellum: 5.9 nTPM (limit 2); cerebral cortex: 6 nTPM (limit 2); heart muscle: 4.8 nTPM (limit 2); hippocampal formation: 2.6 nTPM (limit 2); hypothalamus: 5.9 nTPM (limit 2); midbrain: 2.6 nTPM (limit 2); pituitary gland: 9.7 nTPM ... [full text in table]

Cancer RNA. Kidney Chromophobe (TCGA): 24/64 (37.5%); Lung Adenocarcinoma (validation): 6/105 (5.7%); Testicular Germ Cell Tumor (TCGA): 7/133 (5.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. E3 ubiquitin-protein ligase that mediates polyubiquitination of CD4. E3 ubiquitin ligases accept ubiquitin from an E2 ubiquitin-conjugating enzyme in the form of a thioester and then directly transfer the ubiquitin to targeted substrates. May play a role in ubuquitin-dependent protein sorting in developmenting spermatids

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NNE9 | HPA portal (live; not the pinned values)

MATCAP2 | Unsafe / RNA

ENSG00000164542

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 7.8 nTPM (limit 2); amygdala: 3.6 nTPM (limit 2); basal ganglia: 4.6 nTPM (limit 2); bone marrow: 2.4 nTPM (limit 2); cerebellum: 17.4 nTPM (limit 2); cerebral cortex: 5 nTPM (limit 2); choroid plexus: 6.4 nTPM (limit 2); hippocampal formation: 3.7 nTPM (limit 2); hypothalamus: 5.7 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Pancreatic Adenocarcinoma (validation): 71/80 (88.8%)

Cancer IHC. glioma: 10/12 (83.3%); breast cancer: 9/12 (75.0%)

Biology. Putative tyrosine carboxypeptidase

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCT3 | HPA portal (live; not the pinned values)

MBD1 | Unsafe / Protein

ENSG00000141644

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%)

Cancer IHC. pancreatic cancer: 8/11 (72.7%); breast cancer: 8/12 (66.7%)

Biology. Transcriptional repressor that binds CpG islands in promoters where the DNA is methylated at position 5 of cytosine within CpG dinucleotides. Binding is abolished by the presence of 7-mG that is produced by DNA damage by methylmethanesulfonate (MMS). Acts as transcriptional repressor and plays a role in gene silencing by recruiting ATF7IP, which in turn recruits factors such as the histone methyltransferase SETDB1. Probably forms a ... [full text in table]

Curated tissue note. Widely expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UIS9 | HPA portal (live; not the pinned values)

MBD2 | Unsafe / Protein

ENSG00000134046

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / Purkinje cells: High (Approved); ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Colon Adenocarcinoma (validation): 486/486 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%)

Cancer IHC. urothelial cancer: 9/11 (81.8%); lung cancer: 6/12 (50.0%)

Biology. Binds CpG islands in promoters where the DNA is methylated at position 5 of cytosine within CpG dinucleotides (PubMed:9774669). Binds hemimethylated DNA as well (PubMed:10947852, PubMed:24307175). Recruits histone deacetylases and DNA methyltransferases to chromatin (PubMed:10471499, PubMed:10947852). Acts as a component of the histone deacetylase NuRD complex which participates in the remodeling of chromatin (PubMed:16428440, ... [full text in table]

Curated tissue note. Highly expressed in brain, heart, kidney, stomach, testis and placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UBB5 | HPA portal (live; not the pinned values)

MBD3 | Unsafe / Protein

ENSG00000071655

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Supported); adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: Low (Supported); caudate / glial cells: Low (Supported); caudate / neuronal cells: Low (Supported); ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Ovary Serous Cystadenocarcinoma (validation): 81/81 (100.0%)

Cancer IHC. colorectal cancer: 11/12 (91.7%); glioma: 11/12 (91.7%)

Biology. Acts as a component of the histone deacetylase NuRD complex which participates in the remodeling of chromatin (PubMed:12124384, PubMed:16428440, PubMed:28977666). Acts as transcriptional repressor and plays a role in gene silencing (PubMed:10947852, PubMed:18644863). Does not bind to methylated DNA by itself (PubMed:12124384, PubMed:16428440). Binds to a lesser degree DNA containing unmethylated CpG dinucleotides (PubMed:24307175). ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95983 | HPA portal (live; not the pinned values)

MBD3L1 | Unsafe / Protein

ENSG00000170948

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / proximal tubules (cell body): Medium (Supported)

Cancer RNA. Kidney Chromophobe (TCGA): 1/64 (1.6%); Ovary Serous Cystadenocarcinoma (validation): 1/81 (1.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 2/176 (1.1%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Transcriptional repressor

Curated tissue note. Highly expressed in testis. Detected at low levels in pancreas. Not detected in the other tissues tested

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WWY6 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

MC3R | Unsafe / Protein

ENSG00000124089

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. hypothalamus / neuronal cells: Medium (Uncertain); hypothalamus / neuronal projections: Low (Uncertain); hypothalamus / synapses: Medium (Uncertain)

Cancer RNA. Kidney Chromophobe (TCGA): 1/64 (1.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Lung Squamous Cell Carcinoma (TCGA): 1/489 (0.2%)

Cancer IHC. colorectal cancer: 6/12 (50.0%); stomach cancer: 5/11 (45.5%)

Biology. G protein-coupled receptor for melanocyte-stimulating hormones (alpha, beta, and gamma-MSH) and corticotropin/ACTH, which are peptide products of the POMC precursor (PubMed:37524700, PubMed:8463333). Upon activation, couples to G(s) protein, stimulating adenylate cyclase and the cAMP-dependent signaling pathway, which contributes to the regulation of energy homeostasis (PubMed:18231126, PubMed:37524700, PubMed:8463333). Required for ... [full text in table]

Curated tissue note. Brain, placental, and gut tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P41968 | HPA portal (live; not the pinned values)

MCF2 | Unsafe / RNA

ENSG00000101977

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 23.2 nTPM (limit 2); amygdala: 5.3 nTPM (limit 2); basal ganglia: 7.4 nTPM (limit 2); cerebellum: 3.4 nTPM (limit 2); cerebral cortex: 9 nTPM (limit 2); hippocampal formation: 5.7 nTPM (limit 2); hypothalamus: 11.6 nTPM (limit 2); midbrain: 9.1 nTPM (limit 2); pituitary gland: 3.3 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 15/58 (25.9%); Skin Cuteneous Melanoma (TCGA): 4/99 (4.0%); Kidney Renal Clear Cell Carcinoma (validation): 3/100 (3.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Guanine nucleotide exchange factor (GEF) that modulates the Rho family of GTPases. Promotes the conversion of some member of the Rho family GTPase from the GDP-bound to the GTP-bound form. Isoform 1 exhibits no activity toward RHOA, RAC1 or CDC42. Isoform 2 exhibits decreased GEF activity toward CDC42. Isoform 3 exhibits a weak but significant activity toward RAC1 and CDC42. Isoform 4 exhibits significant activity toward RHOA and ... [full text in table]

Curated tissue note. Isoform 1 is expressed only in brain. Isoform 3 is expressed in heart, kidney, spleen, liver and testis. Isoform 4 is expressed in brain, heart, kidney, testis, placenta, stomach and peripheral blood. The ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P10911 | HPA portal (live; not the pinned values)

MCM10 | Unsafe / Protein

ENSG00000065328

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: High (Approved); lymph node / non-germinal center cells: Medium (Approved); spleen / cells in red pulp: Medium (Approved)

Cancer RNA. Colon Adenocarcinoma (validation): 458/486 (94.2%); Rectum Adenocarcinoma (validation): 178/207 (86.0%); Glioblastoma Multiforme (validation): 47/58 (81.0%)

Cancer IHC. skin cancer: 4/11 (36.4%); cervical cancer: 3/12 (25.0%)

Biology. Acts as a replication initiation factor that brings together the MCM2-7 helicase and the DNA polymerase alpha/primase complex in order to initiate DNA replication. Additionally, plays a role in preventing DNA damage during replication. Key effector of the RBBP6 and ZBTB38-mediated regulation of DNA-replication and common fragile sites stability; acts as a direct target of transcriptional repression by ZBTB38 (PubMed:24726359)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7L590 | HPA portal (live; not the pinned values)

MCMDC2 | Unsafe / RNA

ENSG00000178460

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 4.1 nTPM (limit 2); heart muscle: 2.4 nTPM (limit 2); retina: 4.3 nTPM (limit 2); skeletal muscle: 5.8 nTPM (limit 2); bone marrow: 4.1 nTPM (baseline 2); heart muscle: 2.4 nTPM (baseline 2); retina: 4.3 nTPM (baseline 2); skeletal muscle: 5.8 nTPM (baseline 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 200/349 (57.3%); Stomach Adenocarcinoma (TCGA): 170/346 (49.1%); Rectum Adenocarcinoma (TCGA): 27/88 (30.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays an important role in meiotic recombination and associated DNA double-strand break repair

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4G0Z9 | HPA portal (live; not the pinned values)

MCOLN2 | Unsafe / RNA

ENSG00000153898

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 8.5 nTPM (limit 2); bone marrow: 2.7 nTPM (limit 2); colon: 9.8 nTPM (limit 6); lung: 5.3 nTPM (limit 2); lymph node: 12.5 nTPM (limit 6); rectum: 14.6 nTPM (limit 6); small intestine: 5.3 nTPM (limit 2); adrenal gland: 8.5 nTPM (baseline 2); bone marrow: 2.7 nTPM (baseline 2); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 30/50 (60.0%); Rectum Adenocarcinoma (validation): 121/207 (58.5%); Colon Adenocarcinoma (validation): 268/486 (55.1%)

Cancer IHC. prostate cancer: 9/10 (90.0%); breast cancer: 5/11 (45.5%)

Biology. Nonselective cation channel probably playing a role in the regulation of membrane trafficking events. Acts as a Ca(2+)-permeable cation channel with inwardly rectifying activity (PubMed:19885840, PubMed:19940139). May activate ARF6 and be involved in the trafficking of GPI-anchored cargo proteins to the cell surface via the ARF6-regulated recycling pathway (PubMed:17662026). May play a role in immune processes. In adaptive immunity, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZK6 | HPA portal (live; not the pinned values)

MCPH1 | Unsafe / Protein

ENSG00000147316

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Supported); adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: Medium (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / neuronal cells: Medium (Supported); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 79/80 (98.8%); Colon Adenocarcinoma (validation): 443/486 (91.2%)

Cancer IHC. cervical cancer: 12/12 (100.0%); skin cancer: 12/12 (100.0%)

Biology. Implicated in chromosome condensation and DNA damage induced cellular responses. May play a role in neurogenesis and regulation of the size of the cerebral cortex

Curated tissue note. Expressed in fetal brain, liver and kidney

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEM0 | HPA portal (live; not the pinned values)

MDGA2 | Unsafe / RNA

ENSG00000139915

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 3.5 nTPM (limit 2); retina: 4.3 nTPM (limit 2); cerebral cortex: 3.5 nTPM (baseline 2); retina: 4.3 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 22/58 (37.9%); Skin Cuteneous Melanoma (TCGA): 8/99 (8.1%); Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%)

Cancer IHC. breast cancer: 1/11 (9.1%); glioma: 1/11 (9.1%)

Biology. May be involved in cell-cell interactions

Curated tissue note. Detected in Leydig cells, syncytiotrophoblast, duodenal villi epithelial cells and neutrophils from kidney and cutaneous squamous cell carcinoma (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z553 | HPA portal (live; not the pinned values)

MDH1B | Unsafe / Protein

ENSG00000138400

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Approved); bronchus / ciliated cells (ciliary rootlets): Medium (Approved); bronchus / ciliated cells (tip of cilia): High (Approved); nasopharynx / ciliated cells (cilia axoneme): Medium (Approved); nasopharynx / ciliated cells (tip of cilia): ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 45/58 (77.6%); Breast Invasive Carcinoma (validation): 35/50 (70.0%); Ovary Serous Cystadenocarcinoma (validation): 24/81 (29.6%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5I0G3 | HPA portal (live; not the pinned values)

MED12L | Unsafe / Protein

ENSG00000144893

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); caudate / glial cells: Medium (Enhanced); cerebellum / cells in granular layer: Low (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); cerebral cortex / neuropil: Low (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Testicular Germ Cell Tumor (TCGA): 12/133 (9.0%); Breast Invasive Carcinoma (validation): 2/50 (4.0%)

Cancer IHC. glioma: 6/12 (50.0%); skin cancer: 1/10 (10.0%)

Biology. May be a component of the Mediator complex, a coactivator involved in the regulated transcription of nearly all RNA polymerase II-dependent genes. Mediator functions as a bridge to convey information from gene-specific regulatory proteins to the basal RNA polymerase II transcription machinery. Mediator is recruited to promoters by direct interactions with regulatory proteins and serves as a scaffold for the assembly of a functional ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86YW9 | HPA portal (live; not the pinned values)

MED23 | Unsafe / RNA

ENSG00000112282

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 10.2 nTPM (limit 2); amygdala: 2.4 nTPM (limit 2); basal ganglia: 3.1 nTPM (limit 2); bone marrow: 21.7 nTPM (limit 2); cerebellum: 11.7 nTPM (limit 2); cerebral cortex: 7.1 nTPM (limit 2); choroid plexus: 3.5 nTPM (limit 2); colon: 10.2 nTPM (limit 6); duodenum: 11.1 nTPM (limit 2); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Required for transcriptional activation subsequent to the assembly of the pre-initiation complex (By similarity). Component of the Mediator complex, a coactivator involved in the regulated transcription of nearly all RNA polymerase II-dependent genes. Mediator functions as a bridge to convey information from gene-specific regulatory proteins to the basal RNA polymerase II transcription machinery. Mediator is recruited to promoters by ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9ULK4 | HPA portal (live; not the pinned values)

MED26 | Unsafe / Protein

ENSG00000105085

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Low (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (validation): 206/207 (99.5%); Colon Adenocarcinoma (validation): 483/486 (99.4%); Testicular Germ Cell Tumor (TCGA): 125/133 (94.0%)

Cancer IHC. skin cancer: 1/10 (10.0%); melanoma: 1/11 (9.1%)

Biology. Component of the Mediator complex, a coactivator involved in the regulated transcription of nearly all RNA polymerase II-dependent genes. Mediator functions as a bridge to convey information from gene-specific regulatory proteins to the basal RNA polymerase II transcription machinery. Mediator is recruited to promoters by direct interactions with regulatory proteins and serves as a scaffold for the assembly of a functional ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95402 | HPA portal (live; not the pinned values)

MEIG1 | Unsafe / High RNA

ENSG00000197889

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 47.8 nTPM; lung/ciliated cells; hpa_single_cell_tissue; vital; 6.292 TPM; blood/TFH; dice_tfh; vital

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 11/176 (6.2%); Ovary Serous Cystadenocarcinoma (TCGA): 16/349 (4.6%); Breast Invasive Carcinoma (validation): 2/50 (4.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Essential for spermiogenesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JSS6 | HPA portal (live; not the pinned values)

METTL21A | Unsafe / Protein

ENSG00000144401

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. melanoma: 12/12 (100.0%); breast cancer: 11/11 (100.0%)

Biology. Protein-lysine methyltransferase that selectively trimethylates residues in heat shock protein 70 (HSP70) family members. Contributes to the in vivo trimethylation of Lys residues in HSPA1 and HSPA8. In vitro methylates 'Lys-561' in HSPA1, 'Lys-564' in HSPA2, 'Lys-585' in HSPA5, 'Lys-563' in HSPA6 and 'Lys-561' in HSPA8

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WXB1 | HPA portal (live; not the pinned values)

MEX3A | Unsafe / RNA

ENSG00000254726

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.1 nTPM (limit 2); cerebral cortex: 2.1 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 54/58 (93.1%); Uterine Corpus Endometrial Carcinoma (TCGA): 156/176 (88.6%); Breast Invasive Carcinoma (validation): 43/50 (86.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. RNA binding protein, may be involved in post-transcriptional regulatory mechanisms

Curated tissue note. Highest levels found in fetal brain and testis. Detected also in thymus, salivary gland and uterus

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A1L020 | HPA portal (live; not the pinned values)

MFAP3L | Unsafe / Protein

ENSG00000198948

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); heart muscle / cardiomyocytes: Medium (Enhanced); hippocampus / neuronal cells: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); liver / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 137/141 (97.2%); Kidney Renal Papillary Cell Carcinoma (TCGA): 241/282 (85.5%)

Cancer IHC. liver cancer: 2/10 (20.0%); breast cancer: 0/11 (0.0%)

Biology. May participate in the nuclear signaling of EGFR and MAPK1/ERK2. May a have a role in metastasis

Curated tissue note. Highly expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75121 | HPA portal (live; not the pinned values)

MFSD2B | Unsafe / Protein

ENSG00000205639

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: High (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / Purkinje cells: Medium (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); hippocampus / neuronal cells: Low (Enhanced); spleen / cells in red pulp: Medium (Enhanced)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Liver Hepatocellular Carcinoma (validation): 1/231 (0.4%)

Cancer IHC. testis cancer: 3/10 (30.0%); prostate cancer: 3/12 (25.0%)

Biology. Lipid transporter that specifically mediates export of sphingosine-1-phosphate in red blood cells and platelets (PubMed:29045386). Sphingosine-1-phosphate is a signaling sphingolipid and its export from red blood cells into in the plasma is required for red blood cell morphology (By similarity). Sphingosine-1-phosphate export from platelets is required for platelet aggregation and thrombus formation (By similarity). Mediates the export ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NFX1 | HPA portal (live; not the pinned values)

MFSD6L | Unsafe / RNA

ENSG00000185156

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. duodenum: 2.3 nTPM (limit 2); pancreas: 9.1 nTPM (limit 2); pituitary gland: 4.7 nTPM (limit 2); salivary gland: 6.6 nTPM (limit 6); duodenum: 2.3 nTPM (baseline 2); pancreas: 9.1 nTPM (baseline 2); pituitary gland: 4.7 nTPM (baseline 2); salivary gland: 6.6 nTPM (baseline 6)

Cancer RNA. Thyroid Carcinoma (TCGA): 392/495 (79.2%); Breast Invasive Carcinoma (validation): 26/50 (52.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 71/176 (40.3%)

Cancer IHC. ovarian cancer: 7/11 (63.6%); lung cancer: 6/11 (54.5%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IWD5 | HPA portal (live; not the pinned values)

MGAT4C | Unsafe / Protein

ENSG00000182050

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Approved); colon / glandular cells: Low (Approved); heart muscle / cardiomyocytes: Medium (Approved); hippocampus / neuronal cells: Low (Approved); kidney / cells in glomeruli: Low (Approved); kidney / cells in tubules: Low (Approved); rectum / glandular ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 291/495 (58.8%); Glioblastoma Multiforme (validation): 21/58 (36.2%); Glioblastoma Multiforme (TCGA): 2/141 (1.4%)

Cancer IHC. breast cancer: 1/9 (11.1%); colorectal cancer: 1/9 (11.1%)

Biology. Glycosyltransferase that participates in the transfer of N-acetylglucosamine (GlcNAc) to the core mannose residues of N-linked glycans. Catalyzes the formation of the GlcNAcbeta1-4 branch on the GlcNAcbeta1-2Manalpha1-3 arm of the core structure of N-linked glycans. Essential for the production of tri- and tetra-antennary N-linked sugar chains (By similarity). Does not catalyze the transfer of GlcNAc to the Manalpha1-6 arm to form ... [full text in table]

Curated tissue note. Expressed in heart, adrenal gland, testis, liver, brain and fetal brain. Not expressed in pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UBM8 | HPA portal (live; not the pinned values)

MIA2 | Unsafe / Protein

ENSG00000150527

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: High (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular layer: Medium ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. stomach cancer: 11/11 (100.0%); endometrial cancer: 10/10 (100.0%)

Biology. Plays a role in the transport of cargos that are too large to fit into COPII-coated vesicles and require specific mechanisms to be incorporated into membrane-bound carriers and exported from the endoplasmic reticulum (PubMed:21525241, PubMed:25202031, PubMed:27138255, PubMed:27170179). Plays a role in the secretion of lipoproteins, pre-chylomicrons and pre-VLDLs, by participating in their export from the endoplasmic reticulum ... [full text in table]

Curated tissue note. Highly expressed in liver and weakly in testis. Expression was higher in patients with severe fibrosis or inflammation and chronic hepatitis (PubMed:12586826). Isoform 1 is specifically expressed in lung, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96PC5 | HPA portal (live; not the pinned values)

MICAL2 | Unsafe / Protein

ENSG00000133816

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. testis cancer: 11/11 (100.0%); colorectal cancer: 10/11 (90.9%)

Biology. Methionine monooxygenase that promotes depolymerization of F-actin by mediating oxidation of residues 'Met-44' and 'Met-47' on actin to form methionine-sulfoxide, resulting in actin filament disassembly and preventing repolymerization (PubMed:24440334, PubMed:29343822). Regulates the disassembly of branched actin networks also by oxidizing ARP3B-containing ARP2/3 complexes leading to ARP3B dissociation from the network ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O94851 | HPA portal (live; not the pinned values)

MINDY4 | Unsafe / Protein

ENSG00000106125

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular layer: Medium (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 198/207 (95.7%); Pancreatic Adenocarcinoma (validation): 71/80 (88.8%); Colon Adenocarcinoma (validation): 422/486 (86.8%)

Cancer IHC. renal cancer: 7/12 (58.3%); colorectal cancer: 5/12 (41.7%)

Biology. Probable hydrolase that can remove 'Lys-48'-linked conjugated ubiquitin from proteins

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4G0A6 | HPA portal (live; not the pinned values)

MIPOL1 | Unsafe / Protein

ENSG00000151338

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); adrenal gland / glandular cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in molecular layer: Medium (Enhanced); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 79/80 (98.8%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Lung Adenocarcinoma (validation): 70/105 (66.7%)

Cancer IHC. prostate cancer: 9/9 (100.0%); breast cancer: 10/12 (83.3%)

Biology. Expressed very weakly in heart, liver, skeletal muscle, kidney, pancreas and fetal kidney. Not detected in brain, placenta and lung

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TD10 | HPA portal (live; not the pinned values)

MLANA | Unsafe / Protein

ENSG00000120215

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / pigment epithelial cells: Medium (Enhanced)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 97/99 (98.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 9/176 (5.1%); Pancreatic Adenocarcinoma (validation): 4/80 (5.0%)

Cancer IHC. melanoma: 9/12 (75.0%); breast cancer: 0/11 (0.0%)

Biology. Involved in melanosome biogenesis by ensuring the stability of GPR143. Plays a vital role in the expression, stability, trafficking, and processing of melanocyte protein PMEL, which is critical to the formation of stage II melanosomes

Curated tissue note. Expression is restricted to melanoma and melanocyte cell lines and retina

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q16655 | HPA portal (live; not the pinned values)

MLF1 | Unsafe / Protein

ENSG00000178053

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: High (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / Purkinje cells: Low (Enhanced); nasopharynx / respiratory epithelial cells: High (Enhanced); skeletal muscle / myocytes: Low (Enhanced)

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. endometrial cancer: 3/12 (25.0%); lung cancer: 2/12 (16.7%)

Biology. Involved in lineage commitment of primary hemopoietic progenitors by restricting erythroid formation and enhancing myeloid formation. Interferes with erythropoietin-induced erythroid terminal differentiation by preventing cells from exiting the cell cycle through suppression of CDKN1B/p27Kip1 levels. Suppresses COP1 activity via CSN3 which activates p53 and induces cell cycle arrest. Binds DNA and affects the expression of a number of ... [full text in table]

Curated tissue note. Most abundant in testis, ovary, skeletal muscle, heart, kidney and colon. Low expression in spleen, thymus and peripheral blood leukocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P58340 | HPA portal (live; not the pinned values)

MMP11 | Unsafe / Protein

ENSG00000099953

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); heart muscle / cardiomyocytes: Low (Approved); kidney / cells in tubules: Low (Approved); nasopharynx / respiratory epithelial cells: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 50/50 (100.0%); Breast Invasive Carcinoma (TCGA): 1014/1022 (99.2%); Pancreatic Adenocarcinoma (validation): 79/80 (98.8%)

Cancer IHC. testis cancer: 10/11 (90.9%); prostate cancer: 9/10 (90.0%)

Biology. May play an important role in the progression of epithelial malignancies

Curated tissue note. Specifically expressed in stromal cells of breast carcinomas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P24347 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

MMS22L | Unsafe / RNA

ENSG00000146263

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 7 nTPM (limit 2); retina: 2.3 nTPM (limit 2); bone marrow: 7 nTPM (baseline 2); retina: 2.3 nTPM (baseline 2)

Cancer RNA. Colon Adenocarcinoma (validation): 467/486 (96.1%); Rectum Adenocarcinoma (validation): 193/207 (93.2%); Glioblastoma Multiforme (validation): 53/58 (91.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of the MMS22L-TONSL complex, a complex that promotes homologous recombination-mediated repair of double-strand breaks (DSBs) at stalled or collapsed replication forks (PubMed:21055983, PubMed:21055984, PubMed:21055985, PubMed:21113133, PubMed:26527279, PubMed:27338793, PubMed:29478807). The MMS22L-TONSL complex is required to maintain genome integrity during DNA replication (PubMed:21055983, PubMed:21055984, PubMed:21055985, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZRQ5 | HPA portal (live; not the pinned values)

MND1 | Unsafe / Protein

ENSG00000121211

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Supported); lung / macrophages: Medium (Supported); lymph node / germinal center cells: Medium (Supported); lymph node / non-germinal center cells: Medium (Supported); spleen / cells in red pulp: Medium (Supported); spleen / cells in white pulp: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Colon Adenocarcinoma (TCGA): 236/254 (92.9%); Rectum Adenocarcinoma (TCGA): 80/88 (90.9%)

Cancer IHC. lymphoma: 3/11 (27.3%); breast cancer: 2/9 (22.2%)

Biology. Required for proper homologous chromosome pairing and efficient cross-over and intragenic recombination during meiosis (By similarity). Stimulates both DMC1- and RAD51-mediated homologous strand assimilation, which is required for the resolution of meiotic double-strand breaks

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BWT6 | HPA portal (live; not the pinned values)

MNS1 | Unsafe / RNA

ENSG00000138587

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3 nTPM (limit 2); basal ganglia: 4.3 nTPM (limit 2); bone marrow: 4.5 nTPM (limit 2); cerebellum: 7.5 nTPM (limit 2); cerebral cortex: 2.1 nTPM (limit 2); choroid plexus: 22 nTPM (limit 2); duodenum: 2.2 nTPM (limit 2); heart muscle: 2.4 nTPM (limit 2); hippocampal formation: 4.1 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Kidney Renal Clear Cell Carcinoma (validation): 83/100 (83.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); liver cancer: 12/12 (100.0%)

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia axoneme, which is required for motile cilia beating (PubMed:36191189). May play a role in the control of meiotic division and germ cell differentiation through regulation of pairing and recombination during meiosis. Required for sperm flagella assembly (By similarity). May play a role in the assembly and function of the outer dynein ... [full text in table]

Curated tissue note. Expressed in nasal respiratory epithelium and in the sperm

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEH6 | HPA portal (live; not the pinned values)

MOBP | Unsafe / Protein

ENSG00000168314

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / neuropil: Medium (Enhanced)

Cancer RNA. Glioblastoma Multiforme (TCGA): 71/141 (50.4%); Glioblastoma Multiforme (validation): 27/58 (46.6%); Lung Squamous Cell Carcinoma (validation): 2/68 (2.9%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May play a role in compacting or stabilizing the myelin sheath, possibly by binding the negatively charged acidic phospholipids of the cytoplasmic membrane

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13875 | HPA portal (live; not the pinned values)

MORN3 | Unsafe / RNA

ENSG00000139714

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.1 nTPM (limit 2); basal ganglia: 2.2 nTPM (limit 2); cerebral cortex: 2.4 nTPM (limit 2); choroid plexus: 6 nTPM (limit 2); hippocampal formation: 2.5 nTPM (limit 2); amygdala: 2.1 nTPM (baseline 2); basal ganglia: 2.2 nTPM (baseline 2); cerebral cortex: 2.4 nTPM (baseline 2); choroid ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 37/50 (74.0%); Glioblastoma Multiforme (validation): 29/58 (50.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 47/176 (26.7%)

Cancer IHC. endometrial cancer: 12/12 (100.0%); ovarian cancer: 12/12 (100.0%)

Biology. Assembles a suppression complex (suppresome) by tethering SIRT1 and MDM2 to regulate composite modifications of p53/TP53. Confers both deacetylation-mediated functional inactivation, by SIRT1, and ubiquitination-dependent degradation, by MDM2, of p53/TP53, promoting a proliferative and cell survival behaviors (PubMed:29681526). May play a role in the regulation of spermatogenesis (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PF18 | HPA portal (live; not the pinned values)

MPL | Unsafe / High RNA

ENSG00000117400

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 104.6 nTPM; pbmc/platelets; hpa_single_cell_tissue; vital; 133.7 TPM; blood/Progenitor cell; hpa_immune_monaco; vital

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 9/176 (5.1%); Kidney Renal Papillary Cell Carcinoma (TCGA): 10/282 (3.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 9/283 (3.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Receptor for thrombopoietin that regulates hematopoietic stem cell renewal, megakaryocyte differentiation, and platelet formation. Upon activation by THPO, induces rapid tyrosine phosphorylation and activation of JAK2, providing docking sites for many signaling proteins such as STAT5, SHIP/INPP5D, GRB2, SOS1 and PI3K (PubMed:15899890, PubMed:37633268). In turn, These signaling cascades lead to the proliferation, survival, and ... [full text in table]

Curated tissue note. Expressed at a low level in a large number of cells of hematopoietic origin. Isoform 1 and isoform 2 are always found to be coexpressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P40238 | HPA portal (live; not the pinned values)

MROH8 | Unsafe / Protein

ENSG00000101353

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / glial cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 44/58 (75.9%); Testicular Germ Cell Tumor (TCGA): 12/133 (9.0%); Kidney Chromophobe (TCGA): 4/64 (6.2%)

Cancer IHC. renal cancer: 3/11 (27.3%); prostate cancer: 1/11 (9.1%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

HPA portal (live; not the pinned values)

MRPL30 | Unsafe / Protein

ENSG00000185414

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebral cortex / glial cells: Medium (Approved); cerebral cortex / neuronal cells: Medium (Approved); duodenum / glandular cells: ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. prostate cancer: 10/10 (100.0%); endometrial cancer: 10/12 (83.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TCC3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

MRPS17 | Unsafe / Protein

ENSG00000239789

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / neuronal cells: Medium (Approved); cerebral cortex / neuropil: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%)

Cancer IHC. melanoma: 7/12 (58.3%); endometrial cancer: 6/12 (50.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y2R5 | HPA portal (live; not the pinned values)

MS4A14 | Unsafe / RNA

ENSG00000166928

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 4.7 nTPM (limit 2); hippocampal formation: 2.2 nTPM (limit 2); hypothalamus: 2.1 nTPM (limit 2); midbrain: 2.5 nTPM (limit 2); spinal cord: 4 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 32/50 (64.0%); Glioblastoma Multiforme (validation): 16/58 (27.6%); Breast Invasive Carcinoma (TCGA): 192/1022 (18.8%)

Cancer IHC. ovarian cancer: 2/11 (18.2%); colorectal cancer: 1/11 (9.1%)

Biology. May be involved in signal transduction as a component of a multimeric receptor complex

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96JA4 | HPA portal (live; not the pinned values)

MS4A18 | Unsafe / Protein

ENSG00000214782

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / glandular cells: Low (Approved); duodenum / glandular cells: Medium (Approved); rectum / glandular cells: Low (Approved); small intestine / glandular cells: Medium (Approved)

Cancer RNA. Breast Invasive Carcinoma (TCGA): 11/1022 (1.1%); Pancreatic Adenocarcinoma (TCGA): 1/176 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. liver cancer: 3/12 (25.0%); breast cancer: 1/7 (14.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3C1V0 | HPA portal (live; not the pinned values)

MSANTD4 | Unsafe / Protein

ENSG00000170903

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / ciliated cells (cilia axoneme): High (Approved); bronchus / ciliated cells (ciliary rootlets): High (Approved); bronchus / ciliated cells (tip of cilia): High (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. glioma: 9/12 (75.0%); lung cancer: 1/12 (8.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCY6 | HPA portal (live; not the pinned values)

MSH4 | Unsafe / High RNA

ENSG00000057468

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 37.7 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 3.826 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Glioblastoma Multiforme (validation): 10/58 (17.2%); Colon Adenocarcinoma (validation): 9/486 (1.9%); Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Involved in meiotic recombination. Required for reciprocal recombination and proper segregation of homologous chromosomes at meiosis

Curated tissue note. Highly expressed in testis (PubMed:33437391, PubMed:9299235). Also expressed in the ovary (PubMed:9299235)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O15457 | HPA portal (live; not the pinned values)

MSTN | Unsafe / RNA

ENSG00000138379

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 3 nTPM (limit 2); skeletal muscle: 9.7 nTPM (limit 4); tongue: 14.7 nTPM (limit 6); retina: 3 nTPM (baseline 2); skeletal muscle: 9.7 nTPM (baseline 2); tongue: 14.7 nTPM (baseline 6)

Cancer RNA. Glioblastoma Multiforme (validation): 46/58 (79.3%); Glioblastoma Multiforme (TCGA): 56/141 (39.7%); Breast Invasive Carcinoma (validation): 3/50 (6.0%)

Cancer IHC. breast cancer: 1/11 (9.1%); renal cancer: 1/12 (8.3%)

Biology. Acts specifically as a negative regulator of skeletal muscle growth

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14793 | HPA portal (live; not the pinned values)

MTCL3 | Unsafe / RNA

ENSG00000214338

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 8 nTPM (limit 2); lung: 2.3 nTPM (limit 2); retina: 3.3 nTPM (limit 2); spinal cord: 3.6 nTPM (limit 2); cerebral cortex: 8 nTPM (baseline 2); lung: 2.3 nTPM (baseline 2); retina: 3.3 nTPM (baseline 2); spinal cord: 3.6 nTPM (baseline 2)

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 12/100 (12.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 32/282 (11.3%); Glioblastoma Multiforme (TCGA): 10/141 (7.1%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5TF21 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

MTFR2 | Unsafe / Protein

ENSG00000146410

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: Medium (Approved); duodenum / glandular cells: Low (Approved); lymph node / non-germinal center cells: Low (Approved); rectum / glandular cells: Medium (Approved); small intestine ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 442/486 (90.9%); Rectum Adenocarcinoma (validation): 178/207 (86.0%); Lung Squamous Cell Carcinoma (validation): 56/68 (82.4%)

Cancer IHC. colorectal cancer: 7/10 (70.0%); prostate cancer: 4/10 (40.0%)

Biology. May play a role in mitochondrial aerobic respiration essentially in the testis. Can also promote mitochondrial fission (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6P444 | HPA portal (live; not the pinned values)

MTNAP1 | Unsafe / Protein

ENSG00000141219

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Critical regulator of mitochondrial DNA (mtDNA) abundance (PubMed:37676315). Binds dsDNA throughout the mitochondrial genome without sequence specificity and controls mtDNA copy number by promoting its replication (PubMed:37676315). Also plays important roles in mitochondrial metabolism and cell proliferation (PubMed:37676315)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BSJ5 | HPA portal (live; not the pinned values)

MTNR1B | Unsafe / Protein

ENSG00000134640

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: High (Supported); caudate / neuronal cells: Medium (Supported); cerebellum / cells in molecular layer: Medium (Supported); cerebral cortex / glial cells: Low (Supported); cerebral cortex / neuronal cells: High (Supported); hippocampus / glial cells: High (Supported); retina / ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (TCGA): 16/362 (4.4%); Bladder Urothelial Carcinoma (TCGA): 6/169 (3.6%); Liver Hepatocellular Carcinoma (validation): 5/231 (2.2%)

Cancer IHC. endometrial cancer: 2/12 (16.7%); melanoma: 2/12 (16.7%)

Biology. High affinity receptor for melatonin. Likely to mediate the reproductive and circadian actions of melatonin. The activity of this receptor is mediated by pertussis toxin sensitive G proteins that inhibit adenylate cyclase activity

Curated tissue note. Expressed in retina and less in brain and hippocampus

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P49286 | HPA portal (live; not the pinned values)

MTSS1 | Unsafe / Protein

ENSG00000170873

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / glial cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Liver Hepatocellular Carcinoma (TCGA): 360/362 (99.4%); Pancreatic Adenocarcinoma (validation): 79/80 (98.8%)

Cancer IHC. lung cancer: 12/12 (100.0%); testis cancer: 10/10 (100.0%)

Biology. May be related to cancer progression or tumor metastasis in a variety of organ sites, most likely through an interaction with the actin cytoskeleton

Curated tissue note. Expressed in many tissues, including spleen, thymus, prostate, testis, uterus, colon, and peripheral blood

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43312 | HPA portal (live; not the pinned values)

MYBL1 | Unsafe / Protein

ENSG00000185697

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); cerebellum / Purkinje cells: Medium (Uncertain); cerebral cortex / neuronal cells: Low (Uncertain); colon / glandular cells: Medium (Uncertain); duodenum / glandular cells: Medium (Uncertain); kidney / cells in tubules: Medium (Uncertain); pancreas ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Breast Invasive Carcinoma (TCGA): 341/1022 (33.4%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Transcription factor that specifically recognizes the sequence 5'-YAAC[GT]G-3' (PubMed:7987850, PubMed:8058310). Acts as a master regulator of male meiosis by promoting expression of piRNAs: activates expression of both piRNA precursor RNAs and expression of protein-coding genes involved in piRNA metabolism (By similarity). The piRNA metabolic process mediates the repression of transposable elements during meiosis by forming complexes ... [full text in table]

Curated tissue note. Expressed in a variety of lymphoid and solid tumor lines cultured in vitro

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P10243 | HPA portal (live; not the pinned values)

MYBL2 | Unsafe / Protein

ENSG00000101057

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 283/283 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. pancreatic cancer: 10/11 (90.9%); testis cancer: 9/10 (90.0%)

Biology. Transcription factor involved in the regulation of cell survival, proliferation, and differentiation. Transactivates the expression of the CLU gene

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P10244 | HPA portal (live; not the pinned values)

MYCBPAP | Unsafe / Protein

ENSG00000136449

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Low (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cilia axoneme): Medium (Enhanced); nasopharynx / ciliated cells (tip of cilia): Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 15/50 (30.0%); Glioblastoma Multiforme (validation): 5/58 (8.6%); Uterine Corpus Endometrial Carcinoma (TCGA): 12/176 (6.8%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Plays a crucial role in centrosome-nuclear envelope docking during spermiogenesis, a key step for anchoring the centrosome to the nucleus that is essential for proper sperm flagellum biogenesis. Also required for acrosome biogenesis, manchette organization, and spermatid head morphogenesis, contributing to the complete and functional assembly of the sperm flagellum

Curated tissue note. Expressed specifically in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TBZ2 | HPA portal (live; not the pinned values)

MYH15 | Unsafe / Protein

ENSG00000144821

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebellum / cells in molecular layer: Low (Uncertain); cerebral cortex / glial cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 8/58 (13.8%); Pancreatic Adenocarcinoma (validation): 2/80 (2.5%); Lung Adenocarcinoma (validation): 2/105 (1.9%)

Cancer IHC. glioma: 1/11 (9.1%); urothelial cancer: 1/11 (9.1%)

Biology. Muscle contraction

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y2K3 | HPA portal (live; not the pinned values)

MYO16 | Unsafe / RNA

ENSG00000041515

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.5 nTPM (limit 2); cerebral cortex: 2.5 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 45/58 (77.6%); Glioblastoma Multiforme (TCGA): 12/141 (8.5%); Breast Invasive Carcinoma (validation): 4/50 (8.0%)

Cancer IHC. liver cancer: 2/12 (16.7%); renal cancer: 1/12 (8.3%)

Biology. Myosins are actin-based motor molecules with ATPase activity. Unconventional myosins serve in intracellular movements. Their highly divergent tails are presumed to bind to membranous compartments, which would be moved relative to actin filaments. May be involved in targeting of the catalytic subunit of protein phosphatase 1 during brain development. Activates PI3K and concomitantly recruits the WAVE1 complex to the close vicinity of ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y6X6 | HPA portal (live; not the pinned values)

MYO3A | Unsafe / Protein

ENSG00000095777

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); colon / glandular cells: Low (Enhanced); colon / peripheral nerve/ganglion: Medium (Enhanced); esophagus / squamous epithelial cells: Low (Enhanced); kidney / cells in glomeruli: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); lymph ... [full text in table]

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 41/100 (41.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 91/283 (32.2%); Kidney Renal Clear Cell Carcinoma (TCGA): 105/521 (20.2%)

Cancer IHC. renal cancer: 5/12 (41.7%); melanoma: 4/10 (40.0%)

Biology. Actin-dependent motor protein with a protein kinase activity, playing an essential role in hearing (PubMed:12032315, PubMed:29880844, PubMed:34788109). Probably also plays a role in vision. Required for normal cochlear hair bundle development and hearing. Plays an important role in the early steps of cochlear hair bundle morphogenesis. Influences the number and lengths of stereocilia to be produced and limits the growth of microvilli ... [full text in table]

Curated tissue note. Strongest expression in retina, retinal pigment epithelial cells, cochlea and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEV4 | HPA portal (live; not the pinned values)

MYO3B | Unsafe / Protein

ENSG00000071909

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Approved); cerebral cortex / glial cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); hippocampus / neuronal cells: Low (Approved); lung / macrophages: Low (Approved); parathyroid gland / glandular cells: Low (Approved)

Cancer RNA. Lung Adenocarcinoma (validation): 12/105 (11.4%); Kidney Chromophobe (TCGA): 6/64 (9.4%); Uterine Corpus Endometrial Carcinoma (TCGA): 16/176 (9.1%)

Cancer IHC. renal cancer: 3/11 (27.3%); breast cancer: 2/12 (16.7%)

Biology. Probable actin-based motor with a protein kinase activity. Required for normal cochlear hair bundle development and hearing. Plays an important role in the early steps of cochlear hair bundle morphogenesis. Influences the number and lengths of stereocilia to be produced and limits the growth of microvilli within the forming auditory hair bundles thereby contributing to the architecture of the hair bundle, including its staircase ... [full text in table]

Curated tissue note. Expressed in retina, kidney and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WXR4 | HPA portal (live; not the pinned values)

MYT1L | Unsafe / RNA

ENSG00000186487

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 17.4 nTPM (limit 2); basal ganglia: 28.7 nTPM (limit 2); cerebellum: 36.2 nTPM (limit 2); cerebral cortex: 39.4 nTPM (limit 2); hippocampal formation: 23.4 nTPM (limit 2); hypothalamus: 18.5 nTPM (limit 2); midbrain: 7.3 nTPM (limit 2); pituitary gland: 14.4 nTPM (limit 2); retina: 4.6 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 18/58 (31.0%); Testicular Germ Cell Tumor (TCGA): 13/133 (9.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 16/176 (9.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcription factor that plays a key role in neuronal differentiation by specifically repressing expression of non-neuronal genes during neuron differentiation. In contrast to other transcription repressors that inhibit specific lineages, mediates repression of multiple differentiation programs. Also represses expression of negative regulators of neurogenesis, such as members of the Notch signaling pathway, including HES1. The ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UL68 | HPA portal (live; not the pinned values)

N4BP2 | Unsafe / Protein

ENSG00000078177

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 74/80 (92.5%); Breast Invasive Carcinoma (validation): 38/50 (76.0%)

Cancer IHC. ovarian cancer: 5/12 (41.7%); colorectal cancer: 4/10 (40.0%)

Biology. Has 5'-polynucleotide kinase and nicking endonuclease activity. May play a role in DNA repair or recombination (PubMed:12730195). May play a role in the non-stop mRNA decay pathway (NSD), which specifically targets and degrades mRNAs that lack a proper stop codon (PubMed:40441874)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86UW6 | HPA portal (live; not the pinned values)

NAALADL2 | Unsafe / Protein

ENSG00000177694

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); bronchus / ciliated cells (cell body): Medium (Approved); cerebral cortex / neuronal cells: Low (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: Low (Approved); colon / peripheral nerve/ganglion: Low (Approved); kidney / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 199/207 (96.1%); Pancreatic Adenocarcinoma (validation): 75/80 (93.8%); Glioblastoma Multiforme (validation): 53/58 (91.4%)

Cancer IHC. prostate cancer: 11/11 (100.0%); endometrial cancer: 9/11 (81.8%)

Biology. May be catalytically inactive

Curated tissue note. Expressed at higher level in kidney and placenta. In embryo, it is mainly confined to duodenal and stomach endoderm, mesonephros, metanephros and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q58DX5 | HPA portal (live; not the pinned values)

NACA2 | Unsafe / High RNA

ENSG00000253506

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 289.4 nTPM; bronchus/smooth muscle cells; hpa_single_cell_tissue; vital; 28.34 TPM; blood/CD4_NAIVE; dice_cd4_naive; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 10/50 (20.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 10/283 (3.5%); Uterine Corpus Endometrial Carcinoma (TCGA): 4/176 (2.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Prevents inappropriate targeting of non-secretory polypeptides to the endoplasmic reticulum (ER). Binds to nascent polypeptide chains as they emerge from the ribosome and blocks their interaction with the signal recognition particle (SRP), which normally targets nascent secretory peptides to the ER. Also reduces the inherent affinity of ribosomes for protein translocation sites in the ER membrane (M sites) (By similarity)

Curated tissue note. Expressed specifically in testis and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H009 | HPA portal (live; not the pinned values)

NBPF6 | Unsafe / RNA

ENSG00000186086

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. liver: 2.7 nTPM (limit 2); liver: 2.7 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 31/50 (62.0%); Breast Invasive Carcinoma (TCGA): 231/1022 (22.6%); Rectum Adenocarcinoma (validation): 9/207 (4.3%)

Cancer IHC. melanoma: 6/12 (50.0%); urothelial cancer: 4/12 (33.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VWK0 | HPA portal (live; not the pinned values)

NCAPG2 | Unsafe / Protein

ENSG00000146918

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: High (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. breast cancer: 12/12 (100.0%); ovarian cancer: 12/12 (100.0%)

Biology. Regulatory subunit of the condensin-2 complex, a complex which establishes mitotic chromosome architecture and is involved in physical rigidity of the chromatid axis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86XI2 | HPA portal (live; not the pinned values)

NCAPH | Unsafe / Protein

ENSG00000121152

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); heart muscle / cardiomyocytes: Low (Enhanced); lymph node / germinal center cells: Medium (Enhanced); rectum / glandular cells: Low (Enhanced); skin 1 / cells in basal layer: Low (Enhanced); skin 1 / cells in spinous layer: Low (Enhanced); skin ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 253/254 (99.6%); Colon Adenocarcinoma (validation): 483/486 (99.4%); Testicular Germ Cell Tumor (TCGA): 132/133 (99.2%)

Cancer IHC. lymphoma: 9/12 (75.0%); endometrial cancer: 8/11 (72.7%)

Biology. Regulatory subunit of the condensin complex, a complex required for conversion of interphase chromatin into mitotic-like condense chromosomes. The condensin complex probably introduces positive supercoils into relaxed DNA in the presence of type I topoisomerases and converts nicked DNA into positive knotted forms in the presence of type II topoisomerases (PubMed:11136719). Early in neurogenesis, may play an essential role to ensure ... [full text in table]

Curated tissue note. Widely expressed at low level. Expressed in proliferating cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15003 | HPA portal (live; not the pinned values)

NCKAP5 | Unsafe / RNA

ENSG00000176771

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.6 nTPM (limit 2); basal ganglia: 4.1 nTPM (limit 2); cerebral cortex: 2.5 nTPM (limit 2); duodenum: 2.1 nTPM (limit 2); heart muscle: 2.5 nTPM (limit 2); hippocampal formation: 4.2 nTPM (limit 2); hypothalamus: 2.8 nTPM (limit 2); kidney: 3.9 nTPM (limit 2); liver: 3.9 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 36/50 (72.0%); Lung Squamous Cell Carcinoma (validation): 28/68 (41.2%)

Cancer IHC. liver cancer: 7/12 (58.3%); prostate cancer: 7/12 (58.3%)

Biology. Expressed in fetal and adult brain, leukocytes and fetal fibroblasts

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14513 | HPA portal (live; not the pinned values)

NDUFA11 | Unsafe / RNA

ENSG00000174886

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 192.6 nTPM (limit 10); adrenal gland: 416.4 nTPM (limit 2); amygdala: 296.9 nTPM (limit 2); basal ganglia: 250.5 nTPM (limit 2); bone marrow: 155.9 nTPM (limit 2); cerebellum: 172.8 nTPM (limit 2); cerebral cortex: 266.3 nTPM (limit 2); choroid plexus: 271.5 nTPM (limit 2); colon: ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Accessory subunit of the mitochondrial membrane respiratory chain NADH dehydrogenase (Complex I), that is believed not to be involved in catalysis. Complex I functions in the transfer of electrons from NADH to the respiratory chain. The immediate electron acceptor for the enzyme is believed to be ubiquinone

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86Y39 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

NDUFA2 | Unsafe / Protein

ENSG00000131495

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: High (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. prostate cancer: 10/12 (83.3%); breast cancer: 9/11 (81.8%)

Biology. Accessory subunit of the mitochondrial membrane respiratory chain NADH dehydrogenase (Complex I), that is believed not to be involved in catalysis. Complex I functions in the transfer of electrons from NADH to the respiratory chain. The immediate electron acceptor for the enzyme is believed to be ubiquinone

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43678 | HPA portal (live; not the pinned values)

NDUFA7 | Unsafe / Protein

ENSG00000267855

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: High (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: High (Enhanced); cerebellum / cells in granular layer: High (Enhanced); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); lymphoma: 11/12 (91.7%)

Biology. Accessory subunit of the mitochondrial membrane respiratory chain NADH dehydrogenase (Complex I), that is believed not to be involved in catalysis. Complex I functions in the transfer of electrons from NADH to the respiratory chain. The immediate electron acceptor for the enzyme is believed to be ubiquinone

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95182 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

NEBL | Unsafe / Protein

ENSG00000078114

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); colon / glandular cells: Medium (Approved); duodenum / glandular cells: Medium (Approved); heart muscle / cardiomyocytes: High (Approved); kidney / cells in tubules: Low (Approved); liver / ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 50/50 (100.0%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Rectum Adenocarcinoma (validation): 203/207 (98.1%)

Cancer IHC. pancreatic cancer: 11/12 (91.7%); colorectal cancer: 10/11 (90.9%)

Biology. Binds to actin and plays an important role in the assembly of the Z-disk. May functionally link sarcomeric actin to the desmin intermediate filaments in the heart muscle sarcomeres (PubMed:27733623) May play a role in the assembly of focal adhesions

Curated tissue note. Abundantly expressed in cardiac muscle, but not in skeletal or smooth muscle. Localized to Z-lines in cardiac cells and to dense bodies in nonmuscle cells. Isoform 2 is expressed in non-muscle cells such as in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O76041 | HPA portal (live; not the pinned values)

NEDD1 | Unsafe / Protein

ENSG00000139350

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: High (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); liver cancer: 12/12 (100.0%)

Biology. Required for mitosis progression. Promotes the nucleation of microtubules from the spindle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NHV4 | HPA portal (live; not the pinned values)

NEDD8 | Unsafe / Protein

ENSG00000129559

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. glioma: 11/12 (91.7%); prostate cancer: 11/12 (91.7%)

Biology. Ubiquitin-like protein which plays an important role in cell cycle control and embryogenesis via its conjugation to a limited number of cellular proteins, such as cullins or p53/TP53 (PubMed:10318914, PubMed:10597293, PubMed:11953428, PubMed:14690597, PubMed:15242646, PubMed:9694792, PubMed:38605244, PubMed:38316879). Attachment of NEDD8 to cullins is critical for the recruitment of E2 to the cullin-RING-based E3 ubiquitin-protein ... [full text in table]

Curated tissue note. Highly expressed in heart, skeletal muscle, spleen, thymus, prostate, testis, ovary, colon and leukocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15843 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

NEIL3 | Unsafe / RNA

ENSG00000109674

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 6.5 nTPM (limit 2); thymus: 23 nTPM (limit 10); bone marrow: 6.5 nTPM (baseline 2); thymus: 23 nTPM (baseline 10)

Cancer RNA. Glioblastoma Multiforme (validation): 46/58 (79.3%); Breast Invasive Carcinoma (validation): 34/50 (68.0%); Colon Adenocarcinoma (validation): 292/486 (60.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. DNA glycosylase which prefers single-stranded DNA (ssDNA), or partially ssDNA structures such as bubble and fork structures, to double-stranded DNA (dsDNA) (PubMed:12433996, PubMed:19170771, PubMed:22569481, PubMed:23755964). Mediates interstrand cross-link repair in response to replication stress: acts by mediating DNA glycosylase activity, cleaving one of the two N-glycosyl bonds comprising the interstrand cross-link, which avoids ... [full text in table]

Curated tissue note. Expressed in keratinocytes and embryonic fibroblasts (at protein level). Also detected in thymus, testis and fetal lung primary fibroblasts

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TAT5 | HPA portal (live; not the pinned values)

NEK10 | Unsafe / RNA

ENSG00000163491

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 3.1 nTPM (limit 2); choroid plexus: 2.7 nTPM (limit 2); heart muscle: 3.1 nTPM (limit 2); hippocampal formation: 2.6 nTPM (limit 2); hypothalamus: 2.9 nTPM (limit 2); lung: 2.2 nTPM (limit 2); pituitary gland: 3.3 nTPM (limit 2); retina: 4.6 nTPM (limit 2); skeletal muscle: 9.4 nTPM ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 39/50 (78.0%); Breast Invasive Carcinoma (TCGA): 442/1022 (43.2%); Glioblastoma Multiforme (validation): 17/58 (29.3%)

Cancer IHC. ovarian cancer: 1/11 (9.1%); pancreatic cancer: 1/11 (9.1%)

Biology. Plays a role in the cellular response to UV irradiation. Mediates G2/M cell cycle arrest, MEK autoactivation and ERK1/2-signaling pathway activation in response to UV irradiation. In ciliated cells of airways, it is involved in the regulation of mucociliary transport (PubMed:31959991)

Curated tissue note. Expressed in the lung

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZWH5 | HPA portal (live; not the pinned values)

NEK2 | Unsafe / Protein

ENSG00000117650

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. lymph node / germinal center cells: Medium (Enhanced); lymph node / non-germinal center cells: Medium (Enhanced); rectum / glandular cells: Low (Enhanced); small intestine / glandular cells: Low (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (TCGA): 88/88 (100.0%); Colon Adenocarcinoma (TCGA): 253/254 (99.6%); Colon Adenocarcinoma (validation): 482/486 (99.2%)

Cancer IHC. lymphoma: 5/12 (41.7%); cervical cancer: 2/11 (18.2%)

Biology. Protein kinase which is involved in the control of centrosome separation and bipolar spindle formation in mitotic cells and chromatin condensation in meiotic cells. Regulates centrosome separation (essential for the formation of bipolar spindles and high-fidelity chromosome separation) by phosphorylating centrosomal proteins such as CROCC, CEP250 and NINL, resulting in their displacement from the centrosomes. Regulates kinetochore ... [full text in table]

Curated tissue note. Isoform 1 and isoform 2 are expressed in peripheral blood T-cells and a wide variety of transformed cell types. Isoform 1 and isoform 4 are expressed in the testis. Up-regulated in various cancer cell lines, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P51955 | HPA portal (live; not the pinned values)

NEMP2 | Unsafe / RNA

ENSG00000189362

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 2.3 nTPM (limit 2); cerebellum: 2.3 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 53/58 (91.4%); Pancreatic Adenocarcinoma (validation): 49/80 (61.3%); Colon Adenocarcinoma (validation): 211/486 (43.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NFY4 | HPA portal (live; not the pinned values)

NEUROG2 | Unsafe / RNA

ENSG00000178403

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.1 nTPM (limit 2); basal ganglia: 2.1 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 8/50 (16.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 9/176 (5.1%); Testicular Germ Cell Tumor (TCGA): 5/133 (3.8%)

Cancer IHC. cervical cancer: 12/12 (100.0%); glioma: 12/12 (100.0%)

Biology. Transcriptional regulator. Involved in neuronal differentiation. Activates transcription by binding to the E box (5'-CANNTG-3')

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H2A3 | HPA portal (live; not the pinned values)

NEXMIF | Unsafe / RNA

ENSG00000050030

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 3 nTPM (limit 2); hypothalamus: 2.1 nTPM (limit 2); cerebral cortex: 3 nTPM (baseline 2); hypothalamus: 2.1 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 44/58 (75.9%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 2/283 (0.7%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Involved in neurite outgrowth by regulating cell-cell adhesion via the N-cadherin signaling pathway. May act by regulating expression of protein-coding genes, such as N-cadherins and integrin beta-1 (ITGB1)

Curated tissue note. Highly expressed in fetal and adult brain, predominantly in the cerebral cortex and the cerebellum. Also expressed in other tissues but to a lesser extent

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5QGS0 | HPA portal (live; not the pinned values)

NF1 | Unsafe / Protein

ENSG00000196712

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / cells in molecular layer: Medium (Uncertain); cerebellum / ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. colorectal cancer: 10/10 (100.0%); prostate cancer: 11/12 (91.7%)

Biology. Stimulates the GTPase activity of Ras. NF1 shows greater affinity for Ras GAP, but lower specific activity. May be a regulator of Ras activity

Curated tissue note. Detected in brain, peripheral nerve, lung, colon and muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P21359 | HPA portal (live; not the pinned values)

NHEJ1 | Unsafe / Protein

ENSG00000187736

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Medium (Approved); cerebellum / cells in granular layer: High ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%); Colon Adenocarcinoma (TCGA): 245/254 (96.5%)

Cancer IHC. melanoma: 12/12 (100.0%); cervical cancer: 11/11 (100.0%)

Biology. DNA repair protein involved in DNA non-homologous end joining (NHEJ); it is required for double-strand break (DSB) repair and V(D)J recombination and is also involved in telomere maintenance (PubMed:16439204, PubMed:16439205, PubMed:17317666, PubMed:17470781, PubMed:17717001, PubMed:18158905, PubMed:18644470, PubMed:20558749, PubMed:26100018, PubMed:28369633). Plays a key role in NHEJ by promoting the ligation of various mismatched and ... [full text in table]

Curated tissue note. Ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H9Q4 | HPA portal (live; not the pinned values)

NKAPD1 | Unsafe / Protein

ENSG00000150776

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / ciliated cells (cilia axoneme): Medium (Approved); bronchus / ciliated cells (ciliary rootlets): Medium (Approved); bronchus / ciliated ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. pancreatic cancer: 7/10 (70.0%); prostate cancer: 7/11 (63.6%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZUT1 | HPA portal (live; not the pinned values)

NKAPL | Unsafe / RNA

ENSG00000189134

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.5 nTPM (limit 2); basal ganglia: 2.8 nTPM (limit 2); cerebral cortex: 2.3 nTPM (limit 2); heart muscle: 4.9 nTPM (limit 2); hippocampal formation: 2.3 nTPM (limit 2); hypothalamus: 2.4 nTPM (limit 2); midbrain: 3.6 nTPM (limit 2); retina: 12.2 nTPM (limit 2); skeletal muscle: 5.4 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 10/58 (17.2%); Breast Invasive Carcinoma (validation): 4/50 (8.0%); Kidney Chromophobe (TCGA): 3/64 (4.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcriptional repressor of Notch-mediated signaling. Required for spermatogenesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5M9Q1 | HPA portal (live; not the pinned values)

NKX2-8 | Unsafe / RNA

ENSG00000136327

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. spinal cord: 7.6 nTPM (limit 2); spinal cord: 7.6 nTPM (baseline 2)

Cancer RNA. Thyroid Carcinoma (TCGA): 29/495 (5.9%); Lung Adenocarcinoma (TCGA): 23/497 (4.6%); Lung Squamous Cell Carcinoma (TCGA): 20/489 (4.1%)

Cancer IHC. prostate cancer: 12/12 (100.0%); breast cancer: 10/11 (90.9%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O15522 | HPA portal (live; not the pinned values)

NLGN1 | Unsafe / Protein

ENSG00000169760

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in granular layer: Low (Enhanced); cerebellum / Purkinje cells: Low (Enhanced); cerebral ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 89/100 (89.0%); Glioblastoma Multiforme (TCGA): 121/141 (85.8%)

Cancer IHC. testis cancer: 5/10 (50.0%); colorectal cancer: 5/11 (45.5%)

Biology. Cell surface protein involved in cell-cell-interactions via its interactions with neurexin family members. Plays a role in synapse function and synaptic signal transmission, and probably mediates its effects by recruiting and clustering other synaptic proteins. May promote the initial formation of synapses, but is not essential for this. In vitro, triggers the de novo formation of presynaptic structures. May be involved in ... [full text in table]

Curated tissue note. Expressed in the blood vessel walls (at protein level). Highly expressed in brain through prenatal stages, and at lower levels in pancreas islet beta cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N2Q7 | HPA portal (live; not the pinned values)

NLRP10 | Unsafe / Protein

ENSG00000182261

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 7/492 (1.4%); Uterine Corpus Endometrial Carcinoma (TCGA): 2/176 (1.1%); Skin Cuteneous Melanoma (TCGA): 1/99 (1.0%)

Cancer IHC. melanoma: 10/10 (100.0%); breast cancer: 10/11 (90.9%)

Biology. Inhibits autoprocessing of CASP1, CASP1-dependent IL1B secretion, PYCARD aggregation and PYCARD-mediated apoptosis but not apoptosis induced by FAS or BID (PubMed:15096476). Displays anti-inflammatory activity (PubMed:20393137). Required for immunity against C.albicans infection (By similarity). Involved in the innate immune response by contributing to pro-inflammatory cytokine release in response to invasive bacterial infection ... [full text in table]

Curated tissue note. Highly expressed in basal and suprabasal epidermal cell layers with lower levels in dermal fibroblast cells (at protein level) (PubMed:22672233). Widely expressed with highest levels in heart, brain and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86W26 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

NLRP5 | Unsafe / RNA

ENSG00000171487

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. parathyroid gland: 21.7 nTPM (limit 6); parathyroid gland: 21.7 nTPM (baseline 6)

Cancer RNA. Breast Invasive Carcinoma (validation): 2/50 (4.0%); Breast Invasive Carcinoma (TCGA): 21/1022 (2.1%); Lung Adenocarcinoma (validation): 1/105 (1.0%)

Cancer IHC. colorectal cancer: 9/11 (81.8%); prostate cancer: 8/10 (80.0%)

Biology. Component of the subcortical maternal complex (SCMC), a multiprotein complex that plays a key role in early embryonic development (PubMed:38868925). The SCMC complex is a structural constituent of cytoplasmic lattices, which consist in fibrous structures found in the cytoplasm of oocytes and preimplantation embryos (PubMed:41118754). They are required to store maternal proteins critical for embryonic development, such as ribosomal ... [full text in table]

Curated tissue note. Expressed in cumulus cells (at protein level) (PubMed:19542546). Highly abundant in oocytes and early embryos, however poorly expressed in somatic tissues such as the liver and spinal cord (PubMed:11925379, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P59047 | HPA portal (live; not the pinned values)

NLRP7 | Unsafe / High RNA

ENSG00000167634

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 34.8 nTPM; thymus/plasma cells; hpa_single_cell_tissue; vital; 21.44 TPM; blood/NK; dice_nk; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 45/133 (33.8%); Ovary Serous Cystadenocarcinoma (validation): 11/81 (13.6%); Bladder Urothelial Carcinoma (TCGA): 5/169 (3.0%)

Cancer IHC. liver cancer: 11/11 (100.0%); testis cancer: 11/11 (100.0%)

Biology. Component of cytoplasmic lattices, which consist in fibrous structures found in the cytoplasm of oocytes and preimplantation embryos (PubMed:41118754). Cytoplasmic lattices are required to store maternal proteins critical for embryonic development, such as ribosomal proteins, cytoskeletal tubulins and proteins controlling epigenetic reprogramming of the preimplantation embryo, in a poised but restrained state, regulating their ... [full text in table]

Curated tissue note. Expressed in numerous tissues including uterus and ovary, with low levels in heart and brain. Not detected in skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WX94 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

NME8 | Unsafe / RNA

ENSG00000086288

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 12.2 nTPM (limit 12)

Cancer RNA. Glioblastoma Multiforme (validation): 2/58 (3.4%); Uterine Corpus Endometrial Carcinoma (TCGA): 5/176 (2.8%); Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%)

Cancer IHC. colorectal cancer: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. Possesses an intrinsic kinase activity (PubMed:16313181). In vitro, does not exhibit nucleoside diphosphate kinase (NDPK) activity or disulfide bond-reducing activity (PubMed:11737268, PubMed:16313181). Additionally, exhibits a 3'-5'-DNA exonuclease activity that removes single nucleotides from the 3' terminus of single-stranded DNA substrates and digests overhanging mismatched 3' termini from double-stranded DNA substrates, suggesting ... [full text in table]

Curated tissue note. Testis-specific. Expressed only in primary spermatocytes and round spermatids

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N427 | HPA portal (live; not the pinned values)

NME9 | Unsafe / Protein

ENSG00000181322

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bronchus / ciliated cells (cell body): Low (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Low ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 31/176 (17.6%); Glioblastoma Multiforme (validation): 9/58 (15.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 29/282 (10.3%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May be a regulator of microtubule physiology

Curated tissue note. Detected at very low levels in testis, lung and brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86XW9 | HPA portal (live; not the pinned values)

NOL4 | Unsafe / RNA

ENSG00000101746

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 6.9 nTPM (limit 2); basal ganglia: 8.7 nTPM (limit 2); cerebellum: 10.4 nTPM (limit 2); cerebral cortex: 10 nTPM (limit 2); hippocampal formation: 5.6 nTPM (limit 2); hypothalamus: 7.7 nTPM (limit 2); liver: 2.1 nTPM (limit 2); midbrain: 2.7 nTPM (limit 2); pituitary gland: 7.8 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Glioblastoma Multiforme (TCGA): 46/141 (32.6%); Kidney Renal Papillary Cell Carcinoma (TCGA): 41/282 (14.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Expressed predominantly in fetal brain, adult brain and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O94818 | HPA portal (live; not the pinned values)

NOL4L | Unsafe / Protein

ENSG00000197183

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / cells in granular layer: Low (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Ovary Serous Cystadenocarcinoma (TCGA): 348/349 (99.7%)

Cancer IHC. colorectal cancer: 11/12 (91.7%); pancreatic cancer: 10/11 (90.9%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96MY1 | HPA portal (live; not the pinned values)

NOTUM | Unsafe / RNA

ENSG00000185269

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. liver: 9.7 nTPM (limit 2); lung: 2.1 nTPM (limit 2); skeletal muscle: 7.9 nTPM (limit 2); liver: 9.7 nTPM (baseline 2); lung: 2.1 nTPM (baseline 2); skeletal muscle: 7.9 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (TCGA): 59/88 (67.0%); Rectum Adenocarcinoma (validation): 111/207 (53.6%); Colon Adenocarcinoma (TCGA): 124/254 (48.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Carboxylesterase that acts as a key negative regulator of the Wnt signaling pathway by specifically mediating depalmitoleoylation of WNT proteins. Serine palmitoleoylation of WNT proteins is required for efficient binding to frizzled receptors (PubMed:25731175)

Curated tissue note. Rarely expressed in adult normal tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6P988 | HPA portal (live; not the pinned values)

NOXRED1 | Unsafe / RNA

ENSG00000165555

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 4.2 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (validation): 4/58 (6.9%); Kidney Chromophobe (TCGA): 2/64 (3.1%); Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%)

Cancer IHC. liver cancer: 2/12 (16.7%); breast cancer: 0/12 (0.0%)

Biology. Probable oxidoreductase

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6NXP6 | HPA portal (live; not the pinned values)

NPFFR2 | Unsafe / High RNA

ENSG00000056291

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 194.4 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 0.4645 TPM; blood/NK-cell; hpa_immune_cell_samples; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 8/50 (16.0%); Liver Hepatocellular Carcinoma (validation): 24/231 (10.4%); Bladder Urothelial Carcinoma (TCGA): 11/169 (6.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Receptor for NPAF (A-18-F-amide) and NPFF (F-8-F-amide) neuropeptides, also known as morphine-modulating peptides. Can also be activated by a variety of naturally occurring or synthetic FMRF-amide like ligands. This receptor mediates its action by association with G proteins that activate a phosphatidylinositol-calcium second messenger system

Curated tissue note. Isoform 1 is abundant in placenta. Relatively highly expressed in thymus, testis, and small intestine. Expressed at low levels in several tissues including spleen, prostate, brain, heart, ovary, colon, kidney, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5X5 | HPA portal (live; not the pinned values)

NPHP1 | Unsafe / Protein

ENSG00000144061

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / ciliated cells (ciliary rootlets): High (Approved); caudate / neuronal cells: Low (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / endothelial cells: Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 46/50 (92.0%); Kidney Renal Clear Cell Carcinoma (validation): 90/100 (90.0%); Thyroid Carcinoma (TCGA): 420/495 (84.8%)

Cancer IHC. pancreatic cancer: 10/12 (83.3%); colorectal cancer: 9/12 (75.0%)

Biology. Together with BCAR1 it may play a role in the control of epithelial cell polarity (By similarity). Involved in the organization of apical junctions in kidney cells together with NPHP4 and RPGRIP1L/NPHP8 (By similarity). Does not seem to be strictly required for ciliogenesis (By similarity). Seems to help to recruit PTK2B/PYK2 to cell matrix adhesions, thereby initiating phosphorylation of PTK2B/PYK2 and PTK2B/PYK2-dependent signaling ... [full text in table]

Curated tissue note. Widespread expression, with highest levels in pituitary gland, spinal cord, thyroid gland, testis, skeletal muscle, lymph node and trachea. Weakly expressed in heart, kidney and pancreas. Expressed in nasal ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O15259 | HPA portal (live; not the pinned values)

NR6A1 | Unsafe / RNA

ENSG00000148200

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 4.3 nTPM (limit 2); liver: 2.6 nTPM (limit 2); pancreas: 2.8 nTPM (limit 2); retina: 3 nTPM (limit 2); small intestine: 2.5 nTPM (limit 2); bone marrow: 4.3 nTPM (baseline 2); liver: 2.6 nTPM (baseline 2); pancreas: 2.8 nTPM (baseline 2); retina: 3 nTPM (baseline 2); small intestine: ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 125/133 (94.0%); Rectum Adenocarcinoma (validation): 180/207 (87.0%); Colon Adenocarcinoma (validation): 351/486 (72.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Orphan nuclear receptor that binds to a response element containing the sequence 5'-TCAAGGTCA-3' (PubMed:26769970). Acts as a regulator of embryonic stem cell pluripotency by mediating repression of POU5F1/OCT4: binds to the DR0 element within the POU5F1/OCT4 promoter and inhibits POU5F1/OCT4 expression during embryonic stem cell differentiation (PubMed:26769970). Involved in the regulation of gene expression in germ cell development ... [full text in table]

Curated tissue note. Shows highest expression in the germ cells of the adult testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15406 | HPA portal (live; not the pinned values)

NRG1 | Unsafe / Protein

ENSG00000157168

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebellum / cells in molecular layer: Low (Uncertain); cerebellum / Purkinje cells: Low (Uncertain); cerebral cortex / glial cells: Low (Uncertain); ... [full text in table]

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 404/492 (82.1%); Rectum Adenocarcinoma (validation): 164/207 (79.2%); Kidney Renal Papillary Cell Carcinoma (TCGA): 209/282 (74.1%)

Cancer IHC. lymphoma: 7/12 (58.3%); breast cancer: 0/12 (0.0%)

Biology. Direct ligand for ERBB3 and ERBB4 tyrosine kinase receptors. Concomitantly recruits ERBB1 and ERBB2 coreceptors, resulting in ligand-stimulated tyrosine phosphorylation and activation of the ERBB receptors. The multiple isoforms perform diverse functions such as inducing growth and differentiation of epithelial, glial, neuronal, and skeletal muscle cells; inducing expression of acetylcholine receptor in synaptic vesicles during the ... [full text in table]

Curated tissue note. Type I isoforms are the predominant forms expressed in the endocardium. Isoform alpha is expressed in breast, ovary, testis, prostate, heart, skeletal muscle, lung, placenta liver, kidney, salivary gland, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q02297 | HPA portal (live; not the pinned values)

NRL | Unsafe / Protein

ENSG00000129535

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: High (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 29/50 (58.0%); Kidney Chromophobe (TCGA): 11/64 (17.2%); Testicular Germ Cell Tumor (TCGA): 12/133 (9.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Acts as a transcriptional activator which regulates the expression of several rod-specific genes, including RHO and PDE6B (PubMed:21981118). Also functions as a transcriptional coactivator, stimulating transcription mediated by the transcription factor CRX and NR2E3 (PubMed:17335001). Binds to the rhodopsin promoter in a sequence-specific manner (PubMed:17335001)

Curated tissue note. Expressed in the brain and the retina (PubMed:11477108). Expressed strongly in rod and cone cells (at protein level) (PubMed:11477108)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P54845 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

NSUN3 | Unsafe / RNA

ENSG00000178694

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.7 nTPM (limit 2); basal ganglia: 2.8 nTPM (limit 2); bone marrow: 3.6 nTPM (limit 2); cerebral cortex: 2.1 nTPM (limit 2); duodenum: 3 nTPM (limit 2); heart muscle: 2.3 nTPM (limit 2); hippocampal formation: 2.1 nTPM (limit 2); hypothalamus: 2.2 nTPM (limit 2); kidney: 2.3 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 479/486 (98.6%); Rectum Adenocarcinoma (validation): 204/207 (98.6%)

Cancer IHC. lymphoma: 6/11 (54.5%); prostate cancer: 6/12 (50.0%)

Biology. Mitochondrial tRNA methyltransferase that mediates methylation of cytosine to 5-methylcytosine (m5C) at position 34 of mt-tRNA(Met) (PubMed:27214402, PubMed:27356879, PubMed:27497299). mt-tRNA(Met) methylation at cytosine(34) takes place at the wobble position of the anticodon and initiates the formation of 5-formylcytosine (f(5)c) at this position (PubMed:27214402, PubMed:27356879, PubMed:27497299). mt-tRNA(Met) containing the f(5)c ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H649 | HPA portal (live; not the pinned values)

NSUN7 | Unsafe / Protein

ENSG00000179299

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); cerebral cortex / endothelial cells: Low (Uncertain); colon / glandular cells: Medium (Uncertain); duodenum / glandular cells: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 42/50 (84.0%); Kidney Chromophobe (TCGA): 48/64 (75.0%); Prostate Adenocarcinoma (TCGA): 315/480 (65.6%)

Cancer IHC. melanoma: 5/10 (50.0%); skin cancer: 4/8 (50.0%)

Biology. Plays an essential for sperm flagellum assembly

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NE18 | HPA portal (live; not the pinned values)

NT5DC4 | Unsafe / High RNA

ENSG00000144130

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 6 nTPM; placenta/mixed immune cells; hpa_single_cell_tissue; vital; 137.2 TPM; blood/M2; dice_m2; vital

Cancer RNA. Kidney Chromophobe (TCGA): 2/64 (3.1%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%)

Cancer IHC. colorectal cancer: 9/12 (75.0%); liver cancer: 9/12 (75.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86YG4 | HPA portal (live; not the pinned values)

NUF2 | Unsafe / RNA

ENSG00000143228

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. 90.9 nTPM; tongue/basal keratinocytes; hpa_single_cell_tissue; protected; 9.9 TPM; blood/TREG_MEM; dice_treg_mem; vital

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 67/68 (98.5%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Colon Adenocarcinoma (validation): 473/486 (97.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Acts as a component of the essential kinetochore-associated NDC80 complex, which is required for chromosome segregation and spindle checkpoint activity (PubMed:12438418, PubMed:14654001, PubMed:15062103, PubMed:15235793, PubMed:15239953, PubMed:15548592, PubMed:17535814). Required for kinetochore integrity and the organization of stable microtubule binding sites in the outer plate of the kinetochore (PubMed:15548592). The NDC80 complex ... [full text in table]

Therapeutic evidence. Clinical publication.

UniProt Q9BZD4 | Trial/source record | HPA portal (live; not the pinned values)

NUP58 | Unsafe / Protein

ENSG00000139496

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / neuronal cells: Medium (Approved); colon / glandular cells: Medium (Approved); duodenum / glandular cells: Medium (Approved); esophagus / squamous epithelial cells: Medium (Approved); heart ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. skin cancer: 8/12 (66.7%); lymphoma: 5/11 (45.5%)

Biology. Component of the nuclear pore complex, a complex required for the trafficking across the nuclear membrane

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BVL2 | HPA portal (live; not the pinned values)

NUP62 | Unsafe / Protein

ENSG00000213024

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Supported); adrenal gland / glandular cells: Low (Supported); bone marrow / hematopoietic cells: Medium (Supported); bronchus / ciliated cells (cell body): Medium (Supported); bronchus / ciliated cells (ciliary rootlets): Low (Supported); bronchus / ciliated cells ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%)

Cancer IHC. ovarian cancer: 5/11 (45.5%); renal cancer: 4/12 (33.3%)

Biology. Essential component of the nuclear pore complex (PubMed:1915414). The N-terminal is probably involved in nucleocytoplasmic transport (PubMed:1915414). The C-terminal is involved in protein-protein interaction probably via coiled-coil formation, promotes its association with centrosomes and may function in anchorage of p62 to the pore complex (PubMed:1915414, PubMed:24107630). Plays a role in mitotic cell cycle progression by regulating ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P37198 | HPA portal (live; not the pinned values)

NUP62CL | Unsafe / Protein

ENSG00000198088

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / ciliated cells (cell body): Low (Approved); bronchus / ciliated cells (tip of cilia): Low (Approved); colon / endothelial cells: Low (Approved); colon / peripheral nerve/ganglion: Low (Approved); duodenum / glandular cells: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 409/486 (84.2%); Rectum Adenocarcinoma (validation): 165/207 (79.7%); Breast Invasive Carcinoma (validation): 36/50 (72.0%)

Cancer IHC. melanoma: 9/12 (75.0%); glioma: 8/12 (66.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H1M0 | HPA portal (live; not the pinned values)

NUPR2 | Unsafe / RNA

ENSG00000185290

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.3 nTPM (limit 2); amygdala: 7 nTPM (limit 2); basal ganglia: 6.3 nTPM (limit 2); cerebellum: 6.2 nTPM (limit 2); cerebral cortex: 5 nTPM (limit 2); choroid plexus: 3.4 nTPM (limit 2); hippocampal formation: 2.4 nTPM (limit 2); hypothalamus: 4.9 nTPM (limit 2); kidney: 17.7 nTPM ... [full text in table]

Cancer RNA. Kidney Chromophobe (TCGA): 56/64 (87.5%); Ovary Serous Cystadenocarcinoma (TCGA): 304/349 (87.1%); Thyroid Carcinoma (TCGA): 415/495 (83.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Acts as a transcriptional repressor by inhibiting gene expression at the NUPR1 promoter in a p53/TP53-dependent manner in cancer cells (PubMed:25899918). Involved in the G1 cell cycle arrest, and in a decrease in cell viability and cell proliferation (PubMed:25899918). Plays a role as a negative regulator of the protumoral factor NUPR1 (PubMed:25899918)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NF83 | HPA portal (live; not the pinned values)

NUTM2F | Unsafe / High RNA

ENSG00000130950

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 81.5 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 0.9006 TPM; blood/TFH; dice_tfh; vital

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Ovary Serous Cystadenocarcinoma (TCGA): 1/349 (0.3%); Prostate Adenocarcinoma (TCGA): 1/480 (0.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A1L443 | HPA portal (live; not the pinned values)

NXF2 | Unsafe / RNA

ENSG00000269405

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pituitary gland: 3.2 nTPM (limit 2); pituitary gland: 3.2 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 20/176 (11.4%); Lung Squamous Cell Carcinoma (TCGA): 49/489 (10.0%); Lung Squamous Cell Carcinoma (validation): 6/68 (8.8%)

Cancer IHC. testis cancer: 12/12 (100.0%); colorectal cancer: 10/10 (100.0%)

Biology. Involved in the export of mRNA from the nucleus to the cytoplasm

Curated tissue note. Expressed almost exclusively in testis. Also expressed in several cancers

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9GZY0 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

NXF2B | Unsafe / RNA

ENSG00000269437

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. pituitary gland: 2.4 nTPM (limit 2); pituitary gland: 2.4 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 20/176 (11.4%); Lung Squamous Cell Carcinoma (validation): 5/68 (7.4%); Stomach Adenocarcinoma (TCGA): 25/346 (7.2%)

Cancer IHC. testis cancer: 12/12 (100.0%); colorectal cancer: 10/10 (100.0%)

Biology. Involved in the export of mRNA from the nucleus to the cytoplasm

Curated tissue note. Expressed almost exclusively in testis. Also expressed in several cancers

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9GZY0 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

NXF3 | Unsafe / Protein

ENSG00000147206

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. duodenum / glandular cells: Low (Approved); oral mucosa / squamous epithelial cells: Low (Approved)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 19/80 (23.8%); Pancreatic Adenocarcinoma (TCGA): 34/176 (19.3%); Colon Adenocarcinoma (validation): 55/486 (11.3%)

Cancer IHC. testis cancer: 9/10 (90.0%); urothelial cancer: 6/10 (60.0%)

Biology. May function as a tissue-specific nuclear mRNA export factor

Curated tissue note. Expressed at high level in testis and at low level in a small number of tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H4D5 | HPA portal (live; not the pinned values)

OAZ3 | Unsafe / RNA

ENSG00000143450

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.6 nTPM (limit 2); salivary gland: 7.5 nTPM (limit 6); skeletal muscle: 2.8 nTPM (limit 2); retina: 2.6 nTPM (baseline 2); salivary gland: 7.5 nTPM (baseline 6); skeletal muscle: 2.8 nTPM (baseline 2)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 479/480 (99.8%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Thyroid Carcinoma (TCGA): 440/495 (88.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Ornithine decarboxylase (ODC) antizyme protein that negatively regulates ODC activity and intracellular polyamine biosynthesis and uptake in response to increased intracellular polyamine levels. Binds to ODC monomers, inhibiting the assembly of the functional ODC homodimers. Does not target the ODC monomers for degradation, which allows a protein synthesis-independent restoration of ODC activity (PubMed:17900240). Stabilizes AZIN2 by ... [full text in table]

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UMX2 | HPA portal (live; not the pinned values)

ODAD1 | Unsafe / Protein

ENSG00000105479

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Approved); bronchus / ciliated cells (ciliary rootlets): Low (Approved); bronchus / ciliated cells (tip of cilia): High (Approved); nasopharynx / ciliated cells (ciliary rootlets): High (Approved); nasopharynx / ciliated cells (tip of cilia): High ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 38/50 (76.0%); Ovary Serous Cystadenocarcinoma (validation): 20/81 (24.7%); Uterine Corpus Endometrial Carcinoma (TCGA): 35/176 (19.9%)

Cancer IHC. colorectal cancer: 9/9 (100.0%); breast cancer: 6/11 (54.5%)

Biology. Component of the outer dynein arm-docking complex (ODA-DC) that mediates outer dynein arms (ODA) binding onto the doublet microtubule. Involved in mediating assembly of both ODAs and their axonemal docking complex onto ciliary microtubules (By similarity)

Curated tissue note. Expressed in nasal epithelial cells (PubMed:23261302, PubMed:23261303). Highly expressed in testis and also detected in lung, brain and kidney (PubMed:23261302)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96M63 | HPA portal (live; not the pinned values)

ODAD2 | Unsafe / Protein

ENSG00000169126

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); nasopharynx / ciliated cells (cilia axoneme): Medium (Enhanced); nasopharynx / ciliated cells (ciliary ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 31/176 (17.6%); Breast Invasive Carcinoma (validation): 8/50 (16.0%); Glioblastoma Multiforme (validation): 8/58 (13.8%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Component of the outer dynein arm-docking complex (ODA-DC) that mediates outer dynein arms (ODA) binding onto the doublet microtubule (PubMed:27486780). Involved in mediating assembly of both ODAs and their axonemal docking complex onto ciliary microtubules (PubMed:23849778)

Curated tissue note. Expressed in respiratory epithelial cells (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T2S8 | HPA portal (live; not the pinned values)

ODF2 | Unsafe / Protein

ENSG00000136811

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); lung / macrophages: Medium (Enhanced); nasopharynx / ciliated cells (cilia axoneme): High (Enhanced); nasopharynx / ciliated cells (tip of cilia): High (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Colon Adenocarcinoma (validation): 486/486 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%)

Cancer IHC. colorectal cancer: 5/12 (41.7%); ovarian cancer: 4/11 (36.4%)

Biology. Seems to be a major component of sperm tail outer dense fibers (ODF). ODFs are filamentous structures located on the outside of the axoneme in the midpiece and principal piece of the mammalian sperm tail and may help to maintain the passive elastic structures and elastic recoil of the sperm tail. May have a modulating influence on sperm motility. Functions as a general scaffold protein that is specifically localized at the ... [full text in table]

Curated tissue note. Testis-specific (at protein level). Highly expressed in cytoplasm of step 2 round spermatids. Detected in the middle piece and extends to about half the principal piece of the sperm tails

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5BJF6 | HPA portal (live; not the pinned values)

OIP5 | Unsafe / Protein

ENSG00000104147

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. pancreas / exocrine glandular cells: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 125/133 (94.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 264/283 (93.3%); Breast Invasive Carcinoma (validation): 46/50 (92.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Required for recruitment of CENPA to centromeres and normal chromosome segregation during mitosis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43482 | HPA portal (live; not the pinned values)

OLAH | Unsafe / Protein

ENSG00000152463

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); salivary gland / glandular cells: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 52/133 (39.1%); Thyroid Carcinoma (TCGA): 39/495 (7.9%); Uterine Corpus Endometrial Carcinoma (TCGA): 10/176 (5.7%)

Cancer IHC. liver cancer: 4/12 (33.3%); stomach cancer: 2/9 (22.2%)

Biology. Contributes to the release of free fatty acids from fatty acid synthase (FASN). Has broad substrate specificity, giving rise to a range of free fatty acids with chain lengths between 10 and 16 carbon atoms (C10 - C16)

Curated tissue note. Detected both in lactating and non-lactating breast epithelium (at protein level) (PubMed:6589427). Isoform 2 is up-regulated in bone marrow-derived mononuclear cells of rheumatoid arthritis patients ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NV23 | HPA portal (live; not the pinned values)

ONECUT2 | Unsafe / Protein

ENSG00000119547

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. small intestine / glandular cells: Low (Enhanced)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 196/231 (84.8%); Pancreatic Adenocarcinoma (validation): 60/80 (75.0%); Stomach Adenocarcinoma (TCGA): 151/346 (43.6%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Transcriptional activator. Activates the transcription of a number of liver genes such as HNF3B

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95948 | HPA portal (live; not the pinned values)

OOSP2 | Unsafe / Protein

ENSG00000149507

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); duodenum / glandular cells: Low (Enhanced); heart muscle / cardiomyocytes: Low (Enhanced); skin 1 / keratinocytes: Low (Enhanced); small intestine / glandular cells: Low (Enhanced); stomach 2 / glandular cells: Low (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Involved in oocyte maturation

Curated tissue note. Highly expressed in oocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86WS3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

OPN1LW | Unsafe / RNA

ENSG00000102076

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 248.2 nTPM (limit 2); retina: 248.2 nTPM (baseline 2)

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. G protein-coupled photoreceptor that selectively activates transducin G alpha protein (G(t)) in response to long-wavelength (red) light (PubMed:21314100, PubMed:2937147, PubMed:6140680). This activation leads to increased cGMP phosphodiesterase activity, resulting in decreased intracellular cGMP and subsequent closure of cGMP-gated ion channels (PubMed:21314100, PubMed:2937147, PubMed:6140680). The process is initiated when the ... [full text in table]

Curated tissue note. The red (OPN1LW), green (OPN1MW), and blue (OPN1SW) photoreceptor pigments are found in cone cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P04000 | HPA portal (live; not the pinned values)

OPN5 | Unsafe / Protein

ENSG00000124818

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: High (Supported)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 1/99 (1.0%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. testis cancer: 6/12 (50.0%); lung cancer: 4/12 (33.3%)

Biology. G protein-coupled photoreceptor that selectively activates G(i) protein in response to light in the retina (By similarity). Activation occurs when the opsin-bound cis-retinal chromophore absorbs a photon and isomerizes to all-trans-retinal, inducing a conformational change in the opsin that triggers a G protein-mediated phototransduction cascade (PubMed:22043319). Acts as a bistable protein with absorption peaks at 380 nm (UVA light) ... [full text in table]

Curated tissue note. Detected in brain and retina and cell lines derived from neural retina

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6U736 | HPA portal (live; not the pinned values)

OPRM1 | Unsafe / High RNA

ENSG00000112038

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 71.8 nTPM; brain/inhibitory neurons; hpa_single_cell_tissue; vital; 1.8 TPM; blood/basophil; hpa_immune_monaco; vital

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 8/176 (4.5%); Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%)

Cancer IHC. melanoma: 1/12 (8.3%); breast cancer: 0/10 (0.0%)

Biology. Receptor for endogenous opioids such as beta-endorphin and endomorphin (PubMed:10529478, PubMed:12589820, PubMed:7891175, PubMed:7905839, PubMed:7957926, PubMed:9689128). Receptor for natural and synthetic opioids including morphine, heroin, DAMGO, fentanyl, etorphine, buprenorphin and methadone (PubMed:10529478, PubMed:10836142, PubMed:12589820, PubMed:19300905, PubMed:7891175, PubMed:7905839, PubMed:7957926, PubMed:9689128). Also ... [full text in table]

Curated tissue note. Expressed in brain. Isoform 16 and isoform 17 are detected in brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P35372 | HPA portal (live; not the pinned values)

OR1D2 | Unsafe / RNA

ENSG00000184166

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.3 nTPM (limit 2); retina: 2.3 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 54/207 (26.1%); Colon Adenocarcinoma (validation): 115/486 (23.7%); Testicular Germ Cell Tumor (TCGA): 19/133 (14.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Odorant receptor which may be involved in sperm chemotaxis. Bourgeonal is a strong chemoattractant for sperm in vitro and is shown to be a strong agonist for OR1D2 in vitro. May also function in olfactory reception

Curated tissue note. Expressed in testis. Expressed in spermatozoa (at protein level). Expressed in olfactory epithelium

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P34982 | HPA portal (live; not the pinned values)

OR1J2 | Unsafe / High RNA

ENSG00000197233

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 38 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 4.128 TPM; blood/TH17; dice_th17; vital

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 2/283 (0.7%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Odorant receptor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NGS2 | HPA portal (live; not the pinned values)

OR2A4 | Unsafe / High RNA

ENSG00000180658

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 37.36 nTPM; blood/basophil; hpa_immune_cell_samples; vital; 25.2 TPM; blood/basophil; hpa_immune_monaco; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 45/50 (90.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 104/521 (20.0%); Kidney Renal Clear Cell Carcinoma (validation): 18/100 (18.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Odorant receptor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95047 | HPA portal (live; not the pinned values)

OR3A1 | Unsafe / High RNA

ENSG00000180090

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 32.5 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 3.505 TPM; blood/TREG_NAIVE; dice_treg_naive; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 5/133 (3.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Odorant receptor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P47881 | HPA portal (live; not the pinned values)

OR6C2 | Unsafe / High RNA

ENSG00000179695

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 0.2 nTPM; salivary gland/serous glandular cells; hpa_single_cell_tissue; protected; 30.92 TPM; blood/M2; dice_m2; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Odorant receptor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NZP2 | HPA portal (live; not the pinned values)

OR9A2 | Unsafe / High RNA

ENSG00000179468

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 75.4 nTPM; brain/oligodendrocytes; hpa_single_cell_tissue; vital; 0.812 TPM; blood/TH2; dice_th2; vital

Cancer RNA. Glioblastoma Multiforme (validation): 3/58 (5.2%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Odorant receptor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NGT5 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ORC1 | Unsafe / Protein

ENSG00000085840

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 465/486 (95.7%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 262/283 (92.6%); Testicular Germ Cell Tumor (TCGA): 122/133 (91.7%)

Cancer IHC. renal cancer: 3/12 (25.0%); ovarian cancer: 2/11 (18.2%)

Biology. Component of the origin recognition complex (ORC) that binds origins of replication. DNA-binding is ATP-dependent. The DNA sequences that define origins of replication have not been identified yet. ORC is required to assemble the pre-replication complex necessary to initiate DNA replication

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13415 | HPA portal (live; not the pinned values)

ORC5 | Unsafe / Protein

ENSG00000164815

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. testis cancer: 10/11 (90.9%); urothelial cancer: 8/11 (72.7%)

Biology. Component of the origin recognition complex (ORC) that binds origins of replication. DNA-binding is ATP-dependent. The specific DNA sequences that define origins of replication have not been identified yet. ORC is required to assemble the pre-replication complex necessary to initiate DNA replication

Curated tissue note. Abundant in spleen, ovary, prostate, testis, and colon mucosa

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43913 | HPA portal (live; not the pinned values)

ORC6 | Unsafe / Protein

ENSG00000091651

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: High (Enhanced); cerebellum / Purkinje cells: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); colon / glandular cells: High (Enhanced); duodenum / glandular cells: High (Enhanced); esophagus / squamous epithelial cells: Medium (Enhanced); heart ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Ovary Serous Cystadenocarcinoma (validation): 77/81 (95.1%); Testicular Germ Cell Tumor (TCGA): 116/133 (87.2%)

Cancer IHC. testis cancer: 11/11 (100.0%); cervical cancer: 10/12 (83.3%)

Biology. Component of the origin recognition complex (ORC) that binds origins of replication. DNA-binding is ATP-dependent. The specific DNA sequences that define origins of replication have not been identified yet. ORC is required to assemble the pre-replication complex necessary to initiate DNA replication. Does not bind histone H3 and H4 trimethylation marks H3K9me3, H3K27me3 and H4K20me3

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5N6 | HPA portal (live; not the pinned values)

OTOA | Unsafe / Protein

ENSG00000155719

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. spleen / cells in red pulp: Medium (Enhanced)

Cancer RNA. Kidney Renal Papillary Cell Carcinoma (TCGA): 12/282 (4.3%); Skin Cuteneous Melanoma (TCGA): 3/99 (3.0%); Breast Invasive Carcinoma (validation): 1/50 (2.0%)

Cancer IHC. cervical cancer: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. May act as an adhesion molecule

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7RTW8 | HPA portal (live; not the pinned values)

OTOP2 | Unsafe / RNA

ENSG00000183034

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. colon: 19.4 nTPM (limit 6); rectum: 10.3 nTPM (limit 6); colon: 19.4 nTPM (baseline 6); rectum: 10.3 nTPM (baseline 6)

Cancer RNA. Colon Adenocarcinoma (validation): 18/486 (3.7%); Rectum Adenocarcinoma (TCGA): 2/88 (2.3%); Colon Adenocarcinoma (TCGA): 2/254 (0.8%)

Cancer IHC. colorectal cancer: 6/9 (66.7%); urothelial cancer: 7/11 (63.6%)

Biology. Proton-selective ion channel open at neutral pH. Active at neutral and alkaline extracellular pH, likely participates in some alkali-related physiological activities

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7RTS6 | HPA portal (live; not the pinned values)

OTP | Unsafe / Protein

ENSG00000171540

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. hypothalamus / neuronal cells: Medium (Enhanced)

Cancer RNA. Glioblastoma Multiforme (TCGA): 26/141 (18.4%); Glioblastoma Multiforme (validation): 4/58 (6.9%); Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%)

Cancer IHC. renal cancer: 3/12 (25.0%); liver cancer: 1/10 (10.0%)

Biology. Probably involved in the differentiation of hypothalamic neuroendocrine cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5XKR4 | HPA portal (live; not the pinned values)

OTUD7A | Unsafe / RNA

ENSG00000169918

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 6.1 nTPM (limit 2); basal ganglia: 5.6 nTPM (limit 2); cerebellum: 12 nTPM (limit 2); cerebral cortex: 7.8 nTPM (limit 2); hippocampal formation: 8 nTPM (limit 2); hypothalamus: 5.3 nTPM (limit 2); midbrain: 6.7 nTPM (limit 2); spinal cord: 9.8 nTPM (limit 2); amygdala: 6.1 nTPM (baseline ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 6/58 (10.3%); Kidney Chromophobe (TCGA): 5/64 (7.8%); Breast Invasive Carcinoma (validation): 2/50 (4.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Deubiquitinase, which cleaves 'Lys-11'-linked polyubiquitin chains. Might be required for PA28-20S proteasome assembly (Probable)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TE49 | HPA portal (live; not the pinned values)

OXCT2 | Unsafe / Protein

ENSG00000198754

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / distal tubules: Low (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 13/50 (26.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 25/282 (8.9%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 11/283 (3.9%)

Cancer IHC. colorectal cancer: 2/12 (16.7%); breast cancer: 0/11 (0.0%)

Biology. Key enzyme for ketone body catabolism. Transfers the CoA moiety from succinate to acetoacetate. Formation of the enzyme-CoA intermediate proceeds via an unstable anhydride species formed between the carboxylate groups of the enzyme and substrate (By similarity)

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BYC2 | HPA portal (live; not the pinned values)

PABIR3 | Unsafe / Protein

ENSG00000156500

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); cerebellum / cells in molecular layer: Low (Approved); colon / glandular cells: Medium ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 71/80 (88.8%); Breast Invasive Carcinoma (validation): 39/50 (78.0%); Ovary Serous Cystadenocarcinoma (validation): 63/81 (77.8%)

Cancer IHC. melanoma: 10/11 (90.9%); prostate cancer: 10/11 (90.9%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6P4D5 | HPA portal (live; not the pinned values)

PABPC3 | Unsafe / Protein

ENSG00000151846

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); colon / glandular cells: Low (Approved); duodenum / glandular cells: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 44/50 (88.0%); Glioblastoma Multiforme (validation): 13/58 (22.4%); Uterine Corpus Endometrial Carcinoma (TCGA): 9/176 (5.1%)

Cancer IHC. testis cancer: 11/12 (91.7%); stomach cancer: 8/11 (72.7%)

Biology. Binds the poly(A) tail of mRNA. May be involved in cytoplasmic regulatory processes of mRNA metabolism. Binds poly(A) with a slightly lower affinity as compared to PABPC1

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H361 | HPA portal (live; not the pinned values)

PABPC4L | Unsafe / RNA

ENSG00000254535

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. smooth muscle: 2.3 nTPM (limit 2); smooth muscle: 2.3 nTPM (baseline 2)

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 40/100 (40.0%); Kidney Chromophobe (TCGA): 20/64 (31.2%); Glioblastoma Multiforme (validation): 16/58 (27.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May bind RNA

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P0CB38 | HPA portal (live; not the pinned values)

PAGE2B | Unsafe / High RNA

ENSG00000238269

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 35 nTPM; liver/erythroid cells; hpa_single_cell_tissue; vital; 8.043 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 22/133 (16.5%); Liver Hepatocellular Carcinoma (validation): 30/231 (13.0%); Liver Hepatocellular Carcinoma (TCGA): 39/362 (10.8%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Reconsideration. Review exact loci and source-specific normal-cell signals; reliable protein evidence must address implicated cells.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JRK9 | HPA portal (live; not the pinned values)

PAGE4 | Unsafe / RNA

ENSG00000101951

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. smooth muscle: 23.3 nTPM (limit 12)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 422/480 (87.9%); Liver Hepatocellular Carcinoma (TCGA): 105/362 (29.0%); Liver Hepatocellular Carcinoma (validation): 59/231 (25.5%)

Cancer IHC. liver cancer: 4/9 (44.4%); melanoma: 5/12 (41.7%)

Biology. Intrinsically disordered protein that potentiates the transcriptional activator activity of JUN (PubMed:24263171, PubMed:28289210). Protects cells from stress-induced apoptosis by inhibiting reactive oxygen species (ROS) production and via regulation of the MAPK signaling pathway (PubMed:21357425, PubMed:25374899, PubMed:30658679)

Curated tissue note. Expressed at basal lvels in the adult normal prostate gland but is highly up-regulated in the fetal prostate and prostate cancer cells (PubMed:12489849, PubMed:24263171, PubMed:24559171, PubMed:25374899). ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O60829 | HPA portal (live; not the pinned values)

PAK6 | Unsafe / Protein

ENSG00000137843

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / endothelial cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); cerebral cortex / neuropil: Low (Approved); esophagus / squamous epithelial cells: Medium (Approved); heart muscle / cardiomyocytes: Low (Approved); kidney / cells in tubules: Low (Approved); skin ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 477/495 (96.4%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 272/283 (96.1%); Head and Neck Squamous Cell Carcinoma (TCGA): 458/492 (93.1%)

Cancer IHC. pancreatic cancer: 5/12 (41.7%); skin cancer: 4/12 (33.3%)

Biology. Serine/threonine protein kinase that plays a role in the regulation of gene transcription. The kinase activity is induced by various effectors including AR or MAP2K6/MAPKK6. Phosphorylates the DNA-binding domain of androgen receptor/AR and thereby inhibits AR-mediated transcription. Also inhibits ESR1-mediated transcription. May play a role in cytoskeleton regulation by interacting with IQGAP1. May protect cells from apoptosis through ... [full text in table]

Curated tissue note. Selectively expressed in brain and testis, with lower levels in multiple tissues including prostate and breast

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NQU5 | HPA portal (live; not the pinned values)

PAOX | Unsafe / RNA

ENSG00000148832

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.2 nTPM (limit 2); amygdala: 2.4 nTPM (limit 2); basal ganglia: 2.4 nTPM (limit 2); cerebellum: 2.9 nTPM (limit 2); cerebral cortex: 2.6 nTPM (limit 2); esophagus: 6.1 nTPM (limit 6); hippocampal formation: 2.8 nTPM (limit 2); kidney: 3.3 nTPM (limit 2); liver: 7.5 nTPM (limit 2); ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 213/231 (92.2%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Kidney Renal Clear Cell Carcinoma (validation): 74/100 (74.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Flavoenzyme which catalyzes the oxidation of N(1)-acetylspermine to spermidine and is thus involved in the polyamine back-conversion (PubMed:12477380). Can also oxidize N(1)-acetylspermidine to putrescine. Substrate specificity: N(1)-acetylspermine = N(1)-acetylspermidine > N(1),N(12)-diacylspermine >> spermine. Does not oxidize spermidine. Plays an important role in the regulation of polyamine intracellular concentration and has the ... [full text in table]

Curated tissue note. Widely expressed. Not detected in spleen. Expressed at lower level in neoplastic tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6QHF9 | HPA portal (live; not the pinned values)

PAPOLB | Unsafe / High RNA

ENSG00000218823

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 30.4 nTPM; bronchus/smooth muscle cells; hpa_single_cell_tissue; vital; 0.3377 TPM; blood/CD8_STIM; dice_cd8_stim; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NRJ5 | HPA portal (live; not the pinned values)

PAPPA | Unsafe / Normal

ENSG00000182752

Decision. Published protein production in non-permitted normal cells.

Normal RNA / IHC. 277.6 nTPM; esophagus/fibroblasts; hpa_single_cell_tissue; protected; 1.04 TPM; blood/TREG_MEM; dice_treg_mem; vital

Cancer RNA. Rectum Adenocarcinoma (validation): 93/207 (44.9%); Pancreatic Adenocarcinoma (validation): 33/80 (41.2%); Colon Adenocarcinoma (validation): 177/486 (36.4%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Metalloproteinase which specifically cleaves IGFBP-4 and IGFBP-5, resulting in release of bound IGF. Cleavage of IGFBP-4 is dramatically enhanced by the presence of IGF, whereas cleavage of IGFBP-5 is slightly inhibited by the presence of IGF

Curated tissue note. High levels in placenta and pregnancy serum. In placenta, expressed in X cells in septa and anchoring villi, and in syncytiotrophoblasts in the chorionic villi. Lower levels are found in a variety of other ... [full text in table]

Therapeutic evidence. Normal function.

UniProt Q13219 | Reviewed source 1 | HPA portal (live; not the pinned values)

PAPPA2 | Unsafe / RNA

ENSG00000116183

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 3.6 nTPM (limit 2)

Cancer RNA. Kidney Chromophobe (TCGA): 33/64 (51.6%); Glioblastoma Multiforme (validation): 21/58 (36.2%); Pancreatic Adenocarcinoma (validation): 6/80 (7.5%)

Cancer IHC. breast cancer: 0/9 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Metalloproteinase which specifically cleaves insulin-like growth factor binding protein (IGFBP)-5 at the '163-Ser-|-Lys-164' bond. Shows limited proteolysis toward IGFBP-3

Curated tissue note. Expressed abundantly in placenta, and non-pregnant mammary gland with low expression in the kidney, fetal brain and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BXP8 | HPA portal (live; not the pinned values)

PARPBP | Unsafe / Protein

ENSG00000185480

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 53/58 (91.4%); Colon Adenocarcinoma (validation): 427/486 (87.9%); Lung Squamous Cell Carcinoma (validation): 55/68 (80.9%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Required to suppress inappropriate homologous recombination, thereby playing a central role DNA repair and in the maintenance of genomic stability. Antagonizes homologous recombination by interfering with the formation of the RAD51-DNA homologous recombination structure. Binds single-strand DNA and poly(A) homopolymers. Positively regulate the poly(ADP-ribosyl)ation activity of PARP1; however such function may be indirect

Curated tissue note. Restricted to testis. Overexpressed in multiple cancer cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NWS1 | HPA portal (live; not the pinned values)

PBK | Unsafe / Protein

ENSG00000168078

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. lymph node / germinal center cells: Low (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Colon Adenocarcinoma (validation): 474/486 (97.5%); Lung Squamous Cell Carcinoma (validation): 66/68 (97.1%)

Cancer IHC. lymphoma: 1/11 (9.1%); breast cancer: 0/11 (0.0%)

Biology. Phosphorylates MAP kinase p38. Seems to be active only in mitosis. May also play a role in the activation of lymphoid cells. When phosphorylated, forms a complex with TP53, leading to TP53 destabilization and attenuation of G2/M checkpoint during doxorubicin-induced DNA damage

Curated tissue note. Expressed in the testis and placenta. In the testis, restrictedly expressed in outer cell layer of seminiferous tubules

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96KB5 | HPA portal (live; not the pinned values)

PCDH15 | Unsafe / RNA

ENSG00000150275

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 18.4 nTPM (limit 2); amygdala: 6.8 nTPM (limit 2); basal ganglia: 3.5 nTPM (limit 2); cerebral cortex: 6.6 nTPM (limit 2); hippocampal formation: 5.9 nTPM (limit 2); hypothalamus: 5.1 nTPM (limit 2); kidney: 7.7 nTPM (limit 2); lung: 2.4 nTPM (limit 2); midbrain: 5.1 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 37/58 (63.8%); Glioblastoma Multiforme (TCGA): 37/141 (26.2%); Ovary Serous Cystadenocarcinoma (TCGA): 20/349 (5.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Calcium-dependent cell-adhesion protein. Essential for maintenance of normal retinal and cochlear function

Curated tissue note. Expressed in brain, lung, kidney, spleen and testis. Found also in the inner and outer synaptic layers, and the nerve fiber layer in adult and fetal retinas. Found in the supporting cells, outer sulcus cells ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96QU1 | HPA portal (live; not the pinned values)

PCDHA1 | Unsafe / Protein

ENSG00000204970

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); cerebral cortex / neuropil: Low (Enhanced); hippocampus / neuronal cells: Medium (Enhanced)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 33/231 (14.3%); Glioblastoma Multiforme (validation): 6/58 (10.3%); Breast Invasive Carcinoma (validation): 5/50 (10.0%)

Cancer IHC. endometrial cancer: 2/12 (16.7%); urothelial cancer: 1/10 (10.0%)

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5I3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PCDHA10 | Unsafe / RNA

ENSG00000250120

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 2.9 nTPM (limit 2); cerebral cortex: 8 nTPM (limit 2); pancreas: 4 nTPM (limit 2); retina: 2.3 nTPM (limit 2); spinal cord: 2.6 nTPM (limit 2); cerebellum: 2.9 nTPM (baseline 2); cerebral cortex: 8 nTPM (baseline 2); pancreas: 4 nTPM (baseline 2); retina: 2.3 nTPM (baseline 2); spinal ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 15/58 (25.9%); Thyroid Carcinoma (TCGA): 101/495 (20.4%); Glioblastoma Multiforme (TCGA): 27/141 (19.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5I2 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PCDHA2 | Unsafe / RNA

ENSG00000204969

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 2.4 nTPM (limit 2); cerebellum: 2.4 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 10/58 (17.2%); Liver Hepatocellular Carcinoma (validation): 30/231 (13.0%); Breast Invasive Carcinoma (validation): 6/50 (12.0%)

Cancer IHC. melanoma: 9/11 (81.8%); ovarian cancer: 6/11 (54.5%)

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5H9 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PCDHA4 | Unsafe / Protein

ENSG00000204967

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / neuronal cells: Medium (Approved); colon / glandular cells: Low (Approved); duodenum / glandular cells: Low (Approved); hippocampus / neuronal cells: Medium (Approved); kidney ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 19/58 (32.8%); Breast Invasive Carcinoma (validation): 11/50 (22.0%); Glioblastoma Multiforme (TCGA): 23/141 (16.3%)

Cancer IHC. pancreatic cancer: 1/10 (10.0%); glioma: 1/11 (9.1%)

Biology. Calcium-dependent cell-adhesion protein involved in cells self-recognition and non-self discrimination. Thereby, it is involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UN74 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PCDHA5 | Unsafe / Protein

ENSG00000204965

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular layer: Medium (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / endothelial cells: Low (Approved); ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 29/231 (12.6%); Glioblastoma Multiforme (validation): 6/58 (10.3%); Pancreatic Adenocarcinoma (validation): 4/80 (5.0%)

Cancer IHC. glioma: 5/9 (55.6%); colorectal cancer: 2/11 (18.2%)

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5H7 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PCDHA6 | Unsafe / Protein

ENSG00000081842

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Enhanced); cerebellum / cells in molecular layer: Medium (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); cerebral cortex / neuropil: Medium (Enhanced); hippocampus / neuronal cells: Low (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 13/58 (22.4%); Breast Invasive Carcinoma (validation): 8/50 (16.0%); Liver Hepatocellular Carcinoma (validation): 31/231 (13.4%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UN73 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PCDHAC1 | Unsafe / RNA

ENSG00000248383

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. parathyroid gland: 28.8 nTPM (limit 6); parathyroid gland: 28.8 nTPM (baseline 6)

Cancer RNA. Glioblastoma Multiforme (validation): 27/58 (46.6%); Liver Hepatocellular Carcinoma (validation): 28/231 (12.1%); Pancreatic Adenocarcinoma (validation): 8/80 (10.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H158 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PCDHB2 | Unsafe / Protein

ENSG00000112852

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: Low (Enhanced); cerebellum / cells in molecular layer: Medium (Enhanced); cerebral cortex / glial cells: Low (Enhanced); hippocampus / glial cells: Low (Enhanced); hippocampus / neuronal cells: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); lung / macrophages: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 49/58 (84.5%); Breast Invasive Carcinoma (validation): 37/50 (74.0%); Ovary Serous Cystadenocarcinoma (TCGA): 175/349 (50.1%)

Cancer IHC. glioma: 5/12 (41.7%); endometrial cancer: 1/11 (9.1%)

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5E7 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PCDHB5 | Unsafe / Protein

ENSG00000113209

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / Purkinje cells: High (Approved); cerebral cortex / endothelial cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 49/58 (84.5%); Prostate Adenocarcinoma (TCGA): 264/480 (55.0%); Breast Invasive Carcinoma (validation): 25/50 (50.0%)

Cancer IHC. lymphoma: 10/12 (83.3%); glioma: 4/11 (36.4%)

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5E4 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PCDHB9 | Unsafe / Protein

ENSG00000177839

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Glioblastoma Multiforme (TCGA): 103/141 (73.0%); Breast Invasive Carcinoma (validation): 16/50 (32.0%)

Cancer IHC. liver cancer: 10/10 (100.0%); testis cancer: 9/11 (81.8%)

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5E1 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PCDHGA4 | Unsafe / RNA

ENSG00000262576

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.2 nTPM (limit 2); cerebral cortex: 3.3 nTPM (limit 2); basal ganglia: 2.2 nTPM (baseline 2); cerebral cortex: 3.3 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 46/58 (79.3%); Liver Hepatocellular Carcinoma (validation): 170/231 (73.6%); Breast Invasive Carcinoma (validation): 9/50 (18.0%)

Cancer IHC. glioma: 12/12 (100.0%); melanoma: 11/12 (91.7%)

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5G9 | HPA portal (live; not the pinned values)

PCDHGB1 | Unsafe / RNA

ENSG00000254221

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 12.4 nTPM (limit 2); cerebral cortex: 12.4 nTPM (baseline 2)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 175/231 (75.8%); Glioblastoma Multiforme (validation): 41/58 (70.7%); Glioblastoma Multiforme (TCGA): 37/141 (26.2%)

Cancer IHC. glioma: 12/12 (100.0%); melanoma: 11/12 (91.7%)

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5G3 | HPA portal (live; not the pinned values)

PCDHGB2 | Unsafe / RNA

ENSG00000253910

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3 nTPM (limit 2); basal ganglia: 3 nTPM (limit 2); cerebellum: 3.2 nTPM (limit 2); cerebral cortex: 3.6 nTPM (limit 2); hippocampal formation: 2.1 nTPM (limit 2); amygdala: 3 nTPM (baseline 2); basal ganglia: 3 nTPM (baseline 2); cerebellum: 3.2 nTPM (baseline 2); cerebral cortex: 3.6 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Liver Hepatocellular Carcinoma (validation): 171/231 (74.0%); Glioblastoma Multiforme (TCGA): 94/141 (66.7%)

Cancer IHC. glioma: 12/12 (100.0%); melanoma: 11/12 (91.7%)

Biology. Potential calcium-dependent cell-adhesion protein. May be involved in the establishment and maintenance of specific neuronal connections in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y5G2 | HPA portal (live; not the pinned values)

PCGF5 | Unsafe / Protein

ENSG00000180628

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Approved); bronchus / ciliated cells (tip of cilia): Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / neuronal cells: Low (Approved); duodenum / glandular cells: Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%)

Cancer IHC. stomach cancer: 6/10 (60.0%); lung cancer: 6/11 (54.5%)

Biology. Component of a Polycomb group (PcG) multiprotein PRC1-like complex, a complex class required to maintain the transcriptionally repressive state of many genes, including Hox genes, throughout development. PcG PRC1 complex acts via chromatin remodeling and modification of histones; it mediates monoubiquitination of histone H2A 'Lys-119', rendering chromatin heritably changed in its expressibility (PubMed:26151332). Within the PRC1-like ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86SE9 | HPA portal (live; not the pinned values)

PCP2 | Unsafe / RNA

ENSG00000174788

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 3.7 nTPM (limit 2); cerebellum: 116.2 nTPM (limit 2); heart muscle: 2.1 nTPM (limit 2); hypothalamus: 6.6 nTPM (limit 2); kidney: 7.8 nTPM (limit 2); liver: 3.6 nTPM (limit 2); lung: 2.2 nTPM (limit 2); pancreas: 5.2 nTPM (limit 2); retina: 32.3 nTPM (limit 2); salivary gland: 15.8 ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 47/50 (94.0%); Breast Invasive Carcinoma (TCGA): 577/1022 (56.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 125/283 (44.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May function as a cell-type specific modulator for G protein-mediated cell signaling

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IVA1 | HPA portal (live; not the pinned values)

PCSK4 | Unsafe / RNA

ENSG00000115257

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.4 nTPM (limit 2); basal ganglia: 3 nTPM (limit 2); cerebral cortex: 2.8 nTPM (limit 2); hippocampal formation: 2.5 nTPM (limit 2); hypothalamus: 3.5 nTPM (limit 2); kidney: 3.6 nTPM (limit 2); liver: 3.9 nTPM (limit 2); pancreas: 2.7 nTPM (limit 2); pituitary gland: 6.9 nTPM (limit 2); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 42/50 (84.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 111/282 (39.4%); Liver Hepatocellular Carcinoma (validation): 40/231 (17.3%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Proprotein convertase involved in the processing of hormone and other protein precursors at sites comprised of pairs of basic amino acid residues (By similarity). In males, important for ADAM2 processing as well as other acrosomal proteins with roles in fertilization and critical for normal fertilization events such as sperm capacitation, acrosome reaction and binding of sperm to zona pellucida (By similarity). Also plays a role in ... [full text in table]

Curated tissue note. Placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6UW60 | HPA portal (live; not the pinned values)

PCYT1A | Unsafe / Protein

ENSG00000161217

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Catalyzes the key rate-limiting step in the CDP-choline pathway for phosphatidylcholine biosynthesis

Curated tissue note. Brain, placenta, liver, fetal and adult lung

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P49585 | HPA portal (live; not the pinned values)

PDE10A | Unsafe / Protein

ENSG00000112541

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebellum / white matter cells - cytoplasm/membrane: Low (Supported); kidney / proximal tubules (cell body): Medium (Supported)

Cancer RNA. Glioblastoma Multiforme (validation): 40/58 (69.0%); Breast Invasive Carcinoma (validation): 27/50 (54.0%); Kidney Renal Clear Cell Carcinoma (validation): 32/100 (32.0%)

Cancer IHC. colorectal cancer: 2/11 (18.2%); pancreatic cancer: 1/10 (10.0%)

Biology. Plays a role in signal transduction by regulating the intracellular concentration of cyclic nucleotides (PubMed:10373451, PubMed:10393245, PubMed:16330539, PubMed:17389385, PubMed:27058447). Can hydrolyze both cAMP and cGMP, but has higher affinity for cAMP and is more efficient with cAMP as substrate (PubMed:10373451, PubMed:10393245, PubMed:17389385, PubMed:27058447). May play a critical role in regulating cAMP and cGMP levels in the ... [full text in table]

Curated tissue note. Abundant in the putamen and caudate nucleus regions of brain and testis, moderately expressed in the thyroid gland, pituitary gland, thalamus and cerebellum

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y233 | HPA portal (live; not the pinned values)

PDE11A | Unsafe / Protein

ENSG00000128655

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 27/80 (33.8%); Prostate Adenocarcinoma (TCGA): 107/480 (22.3%); Kidney Chromophobe (TCGA): 13/64 (20.3%)

Cancer IHC. skin cancer: 12/12 (100.0%); pancreatic cancer: 11/11 (100.0%)

Biology. Plays a role in signal transduction by regulating the intracellular concentration of cyclic nucleotides cAMP and cGMP (PubMed:10725373, PubMed:10906126, PubMed:11050148, PubMed:16330539). Catalyzes the hydrolysis of both cAMP and cGMP to 5'-AMP and 5'-GMP, respectively (PubMed:10725373, PubMed:10906126, PubMed:11050148)

Curated tissue note. Isoform 1 is present in prostate, pituitary, heart and liver. It is however not present in testis nor in penis, suggesting that weak inhibition by Tadalafil (Cialis) is not relevant (at protein level). Isoform ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HCR9 | HPA portal (live; not the pinned values)

PDE1A | Unsafe / Protein

ENSG00000115252

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 486/495 (98.2%); Kidney Chromophobe (TCGA): 61/64 (95.3%); Breast Invasive Carcinoma (validation): 42/50 (84.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 11/11 (100.0%)

Biology. Calcium/calmodulin-dependent cyclic nucleotide phosphodiesterase with a dual specificity for the second messengers cGMP and cAMP, which are key regulators of many important physiological processes. Has a higher efficiency with cGMP compared to cAMP

Curated tissue note. Several tissues, including brain, kidney, testes and heart

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P54750 | HPA portal (live; not the pinned values)

PDILT | Unsafe / Protein

ENSG00000169340

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. stomach 2 / glandular cells: Low (Approved)

Cancer RNA. Rectum Adenocarcinoma (TCGA): 4/88 (4.5%); Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%); Bladder Urothelial Carcinoma (TCGA): 4/169 (2.4%)

Cancer IHC. melanoma: 4/12 (33.3%); lung cancer: 2/12 (16.7%)

Biology. Probable redox-inactive chaperone involved in spermatogenesis

Curated tissue note. Testis-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N807 | HPA portal (live; not the pinned values)

PDP2 | Unsafe / Protein

ENSG00000172840

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: High ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. ovarian cancer: 12/12 (100.0%); cervical cancer: 11/11 (100.0%)

Biology. Mitochondrial enzyme that catalyzes the dephosphorylation and concomitant reactivation of the alpha subunit of the E1 component of the pyruvate dehydrogenase complex (PDC), thereby stimulating the conversion of pyruvate into acetyl-CoA (By similarity). Acts as a crucial regulator of T cell metabolism and function, with a particular focus on T-helper Th17 (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P2J9 | HPA portal (live; not the pinned values)

PENK | Unsafe / Protein

ENSG00000181195

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); adrenal gland / medullary cells: High (Enhanced); caudate / neuronal cells: Medium (Enhanced)

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 130/489 (26.6%); Breast Invasive Carcinoma (validation): 13/50 (26.0%); Glioblastoma Multiforme (TCGA): 36/141 (25.5%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Neuropeptide that competes with and mimic the effects of opiate drugs. They play a role in a number of physiologic functions, including pain perception and responses to stress Met-enkephalin-Arg-Phe neuropeptide acts as a strong ligand of Mu-type opioid receptor OPRM1. Met-enkephalin-Arg-Phe-binding to OPRM1 in the nucleus accumbens of the brain increases activation of OPRM1, leading to long-term synaptic depression of glutamate ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P01210 | HPA portal (live; not the pinned values)

PEX13 | Unsafe / Protein

ENSG00000162928

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); adrenal gland / glandular cells: High (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: High (Enhanced); cerebellum ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); lung cancer: 12/12 (100.0%)

Biology. Component of the PEX13-PEX14 docking complex, a translocon channel that specifically mediates the import of peroxisomal cargo proteins bound to PEX5 receptor (PubMed:28765278, PubMed:8858165, PubMed:9653144). The PEX13-PEX14 docking complex forms a large import pore which can be opened to a diameter of about 9 nm (By similarity). Mechanistically, PEX5 receptor along with cargo proteins associates with the PEX14 subunit of the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92968 | HPA portal (live; not the pinned values)

PEX5L | Unsafe / Protein

ENSG00000114757

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: High (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / cells in granular layer: Medium (Enhanced); cerebral cortex / glial cells: High (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 32/58 (55.2%); Glioblastoma Multiforme (TCGA): 15/141 (10.6%); Skin Cuteneous Melanoma (TCGA): 7/99 (7.1%)

Cancer IHC. colorectal cancer: 4/12 (33.3%); glioma: 3/11 (27.3%)

Biology. Accessory subunit of hyperpolarization-activated cyclic nucleotide-gated (HCN) channels, regulating their cell-surface expression and cyclic nucleotide dependence

Curated tissue note. Mainly expressed in brain. Also expressed in pancreas, testis and pituitary

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYB4 | HPA portal (live; not the pinned values)

PFN4 | Unsafe / RNA

ENSG00000176732

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.9 nTPM (limit 2); retina: 2.9 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 37/58 (63.8%); Breast Invasive Carcinoma (validation): 13/50 (26.0%); Glioblastoma Multiforme (TCGA): 8/141 (5.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Involved in male fertility. Required for manchette development and acrosome biogenesis during spermiogenesis (By similarity). Binds in vitro to phospholipids, including phosphatidylinositol 3-phosphate (PtdIns(3)P), phosphatidylinositol 4,5-bisphosphate (PtdIns(4,5)P2), phosphatidylinositol 4-phosphate (PtdIns(4)P) and phosphatidic acid (PA) (PubMed:19419568). Contrary to other profilin family members, does not bind to actin in vitro ... [full text in table]

Curated tissue note. Expressed in testis, in germ cells in seminiferous tubules (at protein level) (PubMed:15591451, PubMed:19419568). Prominently expressed in pachytene/diplotene stage spematocytes (PubMed:33438010)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NHR9 | HPA portal (live; not the pinned values)

PGAM5 | Unsafe / Protein

ENSG00000247077

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); adrenal gland / glandular cells: High (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Mitochondrial serine/threonine phosphatase that dephosphorylates various substrates and thus plays a role in different biological processes including cellular senescence or mitophagy (PubMed:24746696, PubMed:32439975). Modulates cellular senescence by regulating mitochondrial dynamics. Mechanistically, participates in mitochondrial fission through dephosphorylating DNM1L/DRP1 (PubMed:32439975). Additionally, dephosphorylates MFN2 in a ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96HS1 | HPA portal (live; not the pinned values)

PGBD4 | Unsafe / RNA

ENSG00000182405

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.7 nTPM (limit 2); retina: 2.3 nTPM (limit 2); skeletal muscle: 5.1 nTPM (limit 2); bone marrow: 2.7 nTPM (baseline 2); retina: 2.3 nTPM (baseline 2); skeletal muscle: 5.1 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 15/176 (8.5%)

Cancer IHC. urothelial cancer: 3/10 (30.0%); colorectal cancer: 2/11 (18.2%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96DM1 | HPA portal (live; not the pinned values)

PHC3 | Unsafe / Protein

ENSG00000173889

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Component of a Polycomb group (PcG) multiprotein PRC1-like complex, a complex class required to maintain the transcriptionally repressive state of many genes, including Hox genes, throughout development. PcG PRC1 complex acts via chromatin remodeling and modification of histones; it mediates monoubiquitination of histone H2A 'Lys-119', rendering chromatin heritably changed in its expressibility

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NDX5 | HPA portal (live; not the pinned values)

PHF7 | Unsafe / RNA

ENSG00000010318

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.7 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 46/50 (92.0%); Glioblastoma Multiforme (validation): 49/58 (84.5%); Testicular Germ Cell Tumor (TCGA): 68/133 (51.1%)

Cancer IHC. colorectal cancer: 7/12 (58.3%); endometrial cancer: 3/11 (27.3%)

Biology. E3 ubiquitin-protein ligase which ubiquitinates histone H3 at 'Lys-14' (By similarity). Required for male fertility, via inhibition of SPOP-mediated BRDT degradation when in the presence of acetylated histone H4 in early condensing spermatids (By similarity). Stabilization of BRDT allows it to facilitate histone removal in early condensing spermatids and promote the progression of histone-to-protamine exchange (By similarity). Promotes ... [full text in table]

Curated tissue note. Highly expressed in Sertoli cells, but not in germ cells in adult testis. Expression in embryonic testis is 30-times lower. Highly expressed in colon, spleen, white blood cells, pancreas, lung, liver, placenta ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BWX1 | HPA portal (live; not the pinned values)

PHOSPHO1 | Unsafe / RNA

ENSG00000173868

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 18.7 nTPM (limit 2); cerebral cortex: 3.8 nTPM (limit 2); duodenum: 2.8 nTPM (limit 2); lung: 2.3 nTPM (limit 2); midbrain: 2.5 nTPM (limit 2); skeletal muscle: 3.3 nTPM (limit 2); small intestine: 2.9 nTPM (limit 2); spleen: 13.8 nTPM (limit 10); bone marrow: 18.7 nTPM (baseline 2); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 110/207 (53.1%); Colon Adenocarcinoma (validation): 185/486 (38.1%); Ovary Serous Cystadenocarcinoma (TCGA): 86/349 (24.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Phosphatase that has a high activity toward phosphoethanolamine (PEA) and phosphocholine (PCho) (PubMed:15175005). Involved in the generation of inorganic phosphate for bone mineralization (By similarity). Acts in a non-redundant manner with PHOSPHO1 in skeletal mineralization: while PHOSPHO1 mediates the initiation of hydroxyapatite crystallization in the matrix vesicles (MVs), ALPL/TNAP catalyzes the spread of hydroxyapatite ... [full text in table]

Curated tissue note. Expressed at sites of mineralization in bone and cartilage. Highly expressed in osteoblast cell line SaOS-2 which produces a mineralized matrix, but not in MG-63 cell line, which do not mineralize

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TCT1 | HPA portal (live; not the pinned values)

PHTF1 | Unsafe / RNA

ENSG00000116793

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 7.8 nTPM (limit 2); amygdala: 5.4 nTPM (limit 2); basal ganglia: 5.9 nTPM (limit 2); bone marrow: 18.5 nTPM (limit 2); cerebellum: 13.9 nTPM (limit 2); cerebral cortex: 7.4 nTPM (limit 2); choroid plexus: 28.2 nTPM (limit 2); duodenum: 3.4 nTPM (limit 2); heart muscle: 5.4 nTPM ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 140/141 (99.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Widely expressed with highest levels in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UMS5 | HPA portal (live; not the pinned values)

PIAS2 | Unsafe / Protein

ENSG00000078043

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); esophagus / squamous epithelial cells: Low (Enhanced); heart muscle / cardiomyocytes: Low (Enhanced); hippocampus / neuronal cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 206/207 (99.5%); Colon Adenocarcinoma (validation): 479/486 (98.6%)

Cancer IHC. glioma: 10/10 (100.0%); melanoma: 8/12 (66.7%)

Biology. Functions as an E3-type small ubiquitin-like modifier (SUMO) ligase, stabilizing the interaction between UBE2I and the substrate, and as a SUMO-tethering factor. Plays a crucial role as a transcriptional coregulator in various cellular pathways, including the STAT pathway, the p53 pathway and the steroid hormone signaling pathway. The effects of this transcriptional coregulation, transactivation or silencing may vary depending upon the ... [full text in table]

Curated tissue note. Mainly expressed in testis. Isoform 3 is expressed predominantly in adult testis, weakly in pancreas, embryonic testis and sperm, and at very low levels in other organs

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75928 | HPA portal (live; not the pinned values)

PIH1D2 | Unsafe / RNA

ENSG00000150773

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.1 nTPM (limit 2); choroid plexus: 9.3 nTPM (limit 2); hypothalamus: 2.1 nTPM (limit 2); kidney: 3.7 nTPM (limit 2); lung: 2.8 nTPM (limit 2); pituitary gland: 3.2 nTPM (limit 2); retina: 12 nTPM (limit 2); basal ganglia: 2.1 nTPM (baseline 2); choroid plexus: 9.3 nTPM (baseline 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 52/58 (89.7%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Prostate Adenocarcinoma (TCGA): 406/480 (84.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WWB5 | HPA portal (live; not the pinned values)

PIK3CG | Unsafe / Protein

ENSG00000105851

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); colon / glandular cells: Low (Enhanced); duodenum / glandular cells: Medium (Enhanced); kidney / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Colon Adenocarcinoma (validation): 451/486 (92.8%); Rectum Adenocarcinoma (validation): 184/207 (88.9%)

Cancer IHC. breast cancer: 9/12 (75.0%); pancreatic cancer: 5/12 (41.7%)

Biology. Phosphoinositide-3-kinase (PI3K) that phosphorylates PtdIns(4,5)P2 (Phosphatidylinositol 4,5-bisphosphate) to generate phosphatidylinositol 3,4,5-trisphosphate (PIP3). PIP3 plays a key role by recruiting PH domain-containing proteins to the membrane, including AKT1 and PDPK1, activating signaling cascades involved in cell growth, survival, proliferation, motility and morphology. Links G protein coupled receptor activation to PIP3 ... [full text in table]

Curated tissue note. Pancreas, skeletal muscle, liver and heart

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P48736 | HPA portal (live; not the pinned values)

PIK3R2 | Unsafe / Protein

ENSG00000105647

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 349/349 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Regulatory subunit of phosphoinositide-3-kinase (PI3K), a kinase that phosphorylates PtdIns(4,5)P2 (Phosphatidylinositol 4,5-bisphosphate) to generate phosphatidylinositol 3,4,5-trisphosphate (PIP3). PIP3 plays a key role by recruiting PH domain-containing proteins to the membrane, including AKT1 and PDPK1, activating signaling cascades involved in cell growth, survival, proliferation, motility and morphology. Binds to activated ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00459 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PIK3R3 | Unsafe / Protein

ENSG00000117461

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (TCGA): 1019/1022 (99.7%); Colon Adenocarcinoma (validation): 477/486 (98.1%)

Cancer IHC. lung cancer: 7/12 (58.3%); melanoma: 7/12 (58.3%)

Biology. Binds to activated (phosphorylated) protein-tyrosine kinases through its SH2 domain and regulates their kinase activity. During insulin stimulation, it also binds to IRS-1

Curated tissue note. Highest levels in brain and testis. Lower levels in adipose tissue, kidney, heart, lung and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92569 | HPA portal (live; not the pinned values)

PIWIL1 | Unsafe / High RNA

ENSG00000125207

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 161.1 nTPM; eye/rod photoreceptor cells; hpa_single_cell_tissue; vital; 0.5123 TPM; blood/TFH; dice_tfh; vital

Cancer RNA. Colon Adenocarcinoma (validation): 70/486 (14.4%); Pancreatic Adenocarcinoma (validation): 6/80 (7.5%); Rectum Adenocarcinoma (validation): 15/207 (7.2%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Endoribonuclease that plays a central role in postnatal germ cells by repressing transposable elements and preventing their mobilization, which is essential for the germline integrity. Acts via the piRNA metabolic process, which mediates the repression of transposable elements during meiosis by forming complexes composed of piRNAs and Piwi proteins and governs the methylation and subsequent repression of transposons. Directly binds ... [full text in table]

Curated tissue note. Expressed in spermatocytes and spermatids. Also detected in prostate cancer (at protein level). Detected in most fetal and adult tissues. Expressed in testes, specifically in germline cells; detected in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96J94 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PIWIL2 | Unsafe / RNA

ENSG00000197181

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. duodenum: 5.8 nTPM (limit 2); pancreas: 2.3 nTPM (limit 2); skeletal muscle: 2.4 nTPM (limit 2); duodenum: 5.8 nTPM (baseline 2); pancreas: 2.3 nTPM (baseline 2); skeletal muscle: 2.4 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 70/133 (52.6%); Pancreatic Adenocarcinoma (validation): 13/80 (16.2%); Head and Neck Squamous Cell Carcinoma (TCGA): 42/492 (8.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Endoribonuclease that plays a central role during spermatogenesis by repressing transposable elements and preventing their mobilization, which is essential for the germline integrity (By similarity). Plays an essential role in meiotic differentiation of spermatocytes, germ cell differentiation and in self-renewal of spermatogonial stem cells (By similarity). Acts via the piRNA metabolic process, which mediates the repression of ... [full text in table]

Curated tissue note. Expressed in adult testis and in most tumors

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TC59 | HPA portal (live; not the pinned values)

PJVK | Unsafe / RNA

ENSG00000204311

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.5 nTPM (limit 2); amygdala: 3.5 nTPM (limit 2); basal ganglia: 4.4 nTPM (limit 2); cerebral cortex: 3.2 nTPM (limit 2); choroid plexus: 5.2 nTPM (limit 2); hippocampal formation: 3.7 nTPM (limit 2); hypothalamus: 3.5 nTPM (limit 2); kidney: 4.1 nTPM (limit 2); liver: 4 nTPM (limit ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 32/58 (55.2%); Kidney Renal Papillary Cell Carcinoma (TCGA): 137/282 (48.6%); Uterine Corpus Endometrial Carcinoma (TCGA): 28/176 (15.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Peroxisome-associated protein required to protect auditory hair cells against noise-induced damage. Acts by regulating noise-induced peroxisome proliferation in auditory hair cells and neurons, and promoting autophagic degradation of damaged peroxisomes (pexophagy). Noise overexposure increases reactive oxygen species (ROS) levels, causing oxidative damage to auditory hair cells and resulting in hearing loss. PJVK acts as a ROS sensor ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q0ZLH3 | HPA portal (live; not the pinned values)

PKD1L1 | Unsafe / RNA

ENSG00000158683

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. heart muscle: 2.6 nTPM (limit 2); skeletal muscle: 2.7 nTPM (limit 2); heart muscle: 2.6 nTPM (baseline 2); skeletal muscle: 2.7 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 51/133 (38.3%); Rectum Adenocarcinoma (validation): 21/207 (10.1%); Colon Adenocarcinoma (TCGA): 20/254 (7.9%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); stomach cancer: 12/12 (100.0%)

Biology. Component of a calcium-permeant ion channel formed by PKD1L2 and PKD1L1 in primary cilia, where it controls cilium calcium concentration, without affecting cytoplasmic calcium concentration, and regulates sonic hedgehog/SHH signaling and GLI2 transcription (PubMed:24336289). The PKD1L1:PKD2L1 channel complex is mechanosensitive only at high pressures and is highly temperature sensitive (PubMed:24336289). Also involved in left/right ... [full text in table]

Curated tissue note. Detected in testis and in fetal and adult heart

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TDX9 | HPA portal (live; not the pinned values)

PKD2L2 | Unsafe / Protein

ENSG00000078795

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / Purkinje cells: Low (Approved); ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 70/231 (30.3%); Kidney Chromophobe (TCGA): 12/64 (18.8%); Prostate Adenocarcinoma (TCGA): 73/480 (15.2%)

Cancer IHC. liver cancer: 10/12 (83.3%); prostate cancer: 9/12 (75.0%)

Biology. Exhibits a lower single conductance but no spontaneous channel activity (PubMed:16883570). May function as a regulator of calcium channels or a channel component involving Ca2(+) homeostasis (By similarity)

Curated tissue note. Expressed only in testis (PubMed:10602361). Expressed also in brain and kidney (PubMed:10756092) Expressed only in transformed lymphoblasts

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NZM6 | HPA portal (live; not the pinned values)

PKMYT1 | Unsafe / RNA

ENSG00000127564

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 27.3 nTPM (limit 2); cerebellum: 3.8 nTPM (limit 2); duodenum: 2.7 nTPM (limit 2); esophagus: 11.2 nTPM (limit 6); lymph node: 7.8 nTPM (limit 6); retina: 16.9 nTPM (limit 2); skeletal muscle: 2.3 nTPM (limit 2); small intestine: 3.9 nTPM (limit 2); bone marrow: 27.3 nTPM (baseline 2); ... [full text in table]

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Acts as a negative regulator of entry into mitosis (G2 to M transition) by phosphorylation of the CDK1 kinase specifically when CDK1 is complexed to cyclins (PubMed:10373560, PubMed:10504341, PubMed:9001210, PubMed:9268380). Mediates phosphorylation of CDK1 predominantly on 'Thr-14'. Also involved in Golgi fragmentation (PubMed:9001210, PubMed:9268380). May be involved in phosphorylation of CDK1 on 'Tyr-15' to a lesser degree, however ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q99640 | HPA portal (live; not the pinned values)

PLAAT1 | Unsafe / RNA

ENSG00000127252

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.4 nTPM (limit 2); amygdala: 8.5 nTPM (limit 2); basal ganglia: 6.4 nTPM (limit 2); cerebral cortex: 6.9 nTPM (limit 2); choroid plexus: 3.5 nTPM (limit 2); esophagus: 6.3 nTPM (limit 6); heart muscle: 9 nTPM (limit 2); hippocampal formation: 6.3 nTPM (limit 2); hypothalamus: 5.9 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Glioblastoma Multiforme (TCGA): 119/141 (84.4%); Lung Squamous Cell Carcinoma (TCGA): 237/489 (48.5%)

Cancer IHC. liver cancer: 9/10 (90.0%); pancreatic cancer: 6/7 (85.7%)

Biology. Exhibits both phospholipase A1/2 and acyltransferase activities (PubMed:21880860, PubMed:26503625). Shows phospholipase A1 (PLA1) and A2 (PLA2) activity, catalyzing the calcium-independent release of fatty acids from the sn-1 or sn-2 position of glycerophospholipids (PubMed:21880860, PubMed:22825852, PubMed:27623847). Shows O-acyltransferase activity, catalyzing the transfer of a fatty acyl group from glycerophospholipid to the ... [full text in table]

Curated tissue note. Abundantly expressed in testis, skeletal muscle, brain, and heart Highly expressed in the testis, skeletal muscle, brain, heart, and thyroid

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HDD0 | HPA portal (live; not the pinned values)

PLAC8L1 | Unsafe / RNA

ENSG00000173261

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. spinal cord: 2.4 nTPM (limit 2); spinal cord: 2.4 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 20/58 (34.5%); Breast Invasive Carcinoma (validation): 8/50 (16.0%); Liver Hepatocellular Carcinoma (validation): 16/231 (6.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A1L4L8 | HPA portal (live; not the pinned values)

PLD6 | Unsafe / Protein

ENSG00000179598

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / endothelial cells: Medium (Approved); cerebral cortex / glial cells: Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 47/50 (94.0%); Prostate Adenocarcinoma (TCGA): 435/480 (90.6%); Glioblastoma Multiforme (validation): 38/58 (65.5%)

Cancer IHC. prostate cancer: 12/12 (100.0%); pancreatic cancer: 10/10 (100.0%)

Biology. Presents phospholipase and nuclease activities, depending on the different physiological conditions (PubMed:17028579, PubMed:21397847, PubMed:28063496). Interaction with Mitoguardin (MIGA1 or MIGA2) affects the dimer conformation, facilitating the lipase activity over the nuclease activity (PubMed:26711011). Plays a key role in mitochondrial fusion and fission via its phospholipase activity (PubMed:17028579, PubMed:24599962, ... [full text in table]

Curated tissue note. Predominantly expressed in testis and ovary, but not limited to gonads (at protein level) (PubMed:17028579, PubMed:21397847). It is also found in brain, heart, pituitary gland, prostate, pancreas, thyroid, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N2A8 | HPA portal (live; not the pinned values)

PLEKHM3 | Unsafe / Protein

ENSG00000178385

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 78/80 (97.5%); Breast Invasive Carcinoma (validation): 39/50 (78.0%)

Cancer IHC. colorectal cancer: 9/12 (75.0%); liver cancer: 8/11 (72.7%)

Biology. Involved in skeletal muscle differentiation. May act as a scaffold protein for AKT1 during muscle differentiation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZWE6 | HPA portal (live; not the pinned values)

PLK1 | Unsafe / Protein

ENSG00000166851

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 283/283 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. liver cancer: 4/12 (33.3%); cervical cancer: 3/12 (25.0%)

Biology. Serine/threonine-protein kinase that performs several important functions throughout M phase of the cell cycle, including the regulation of centrosome maturation and spindle assembly, the removal of cohesins from chromosome arms, the inactivation of anaphase-promoting complex/cyclosome (APC/C) inhibitors, and the regulation of mitotic exit and cytokinesis (PubMed:11202906, PubMed:12207013, PubMed:12447691, PubMed:12524548, ... [full text in table]

Curated tissue note. Placenta and colon

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P53350 | HPA portal (live; not the pinned values)

PLK4 | Unsafe / Protein

ENSG00000142731

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebral cortex / endothelial cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); colon / glandular cells: Medium (Approved); duodenum / glandular cells: Medium ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 482/486 (99.2%); Breast Invasive Carcinoma (validation): 49/50 (98.0%); Glioblastoma Multiforme (validation): 56/58 (96.6%)

Cancer IHC. prostate cancer: 9/12 (75.0%); cervical cancer: 8/12 (66.7%)

Biology. Serine/threonine-protein kinase that plays a central role in centriole duplication. Able to trigger procentriole formation on the surface of the parental centriole cylinder, leading to the recruitment of centriole biogenesis proteins such as SASS6, CPAP, CCP110, CEP135 and gamma-tubulin. When overexpressed, it is able to induce centrosome amplification through the simultaneous generation of multiple procentrioles adjoining each ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00444 | HPA portal (live; not the pinned values)

PNLDC1 | Unsafe / Protein

ENSG00000146453

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebellum / Purkinje cells: Low (Uncertain); cerebral cortex / neuropil: Low (Uncertain); ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 34/133 (25.6%); Lung Squamous Cell Carcinoma (validation): 12/68 (17.6%); Head and Neck Squamous Cell Carcinoma (TCGA): 74/492 (15.0%)

Cancer IHC. renal cancer: 11/12 (91.7%); pancreatic cancer: 4/9 (44.4%)

Biology. 3'-exoribonuclease that has a preference for poly(A) tails of mRNAs, thereby efficiently degrading poly(A) tails (PubMed:27515512). Exonucleolytic degradation of the poly(A) tail is often the first step in the decay of eukaryotic mRNAs and is also used to silence certain maternal mRNAs translationally during oocyte maturation and early embryonic development (PubMed:27515512). May act as a regulator of multipotency in embryonic stem ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NA58 | HPA portal (live; not the pinned values)

PNMA5 | Unsafe / Protein

ENSG00000198883

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Medium (Enhanced); cerebral cortex / neuronal cells: High (Enhanced); hippocampus / neuronal cells: Medium (Enhanced); stomach 1 / glandular cells: Low (Enhanced); stomach 2 / glandular cells: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 46/133 (34.6%); Bladder Urothelial Carcinoma (TCGA): 10/169 (5.9%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 16/283 (5.7%)

Cancer IHC. testis cancer: 3/12 (25.0%); urothelial cancer: 2/12 (16.7%)

Biology. Expressed in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96PV4 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

POLE2 | Unsafe / Protein

ENSG00000100479

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Uncertain); esophagus / squamous epithelial cells: Medium (Uncertain); lymph node / non-germinal center cells: Low (Uncertain); oral mucosa / squamous epithelial cells: Medium (Uncertain); skin 2 / epidermal cells: Medium (Uncertain); urinary bladder / ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 46/50 (92.0%); Colon Adenocarcinoma (validation): 443/486 (91.2%); Glioblastoma Multiforme (validation): 51/58 (87.9%)

Cancer IHC. liver cancer: 6/10 (60.0%); renal cancer: 7/12 (58.3%)

Biology. Accessory component of the DNA polymerase epsilon complex (PubMed:10801849). Participates in DNA repair and in chromosomal DNA replication (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P56282 | HPA portal (live; not the pinned values)

POLH | Unsafe / Protein

ENSG00000170734

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. breast cancer: 10/12 (83.3%); colorectal cancer: 10/12 (83.3%)

Biology. DNA polymerase specifically involved in the DNA repair by translesion synthesis (TLS) (PubMed:10385124, PubMed:11743006, PubMed:16357261, PubMed:20388628, PubMed:24449906, PubMed:24553286, PubMed:38212351). Due to low processivity on both damaged and normal DNA, cooperates with the heterotetrameric (REV3L, REV7, POLD2 and POLD3) POLZ complex for complete bypass of DNA lesions. Inserts one or 2 nucleotide(s) opposite the lesion, the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y253 | HPA portal (live; not the pinned values)

POLQ | Unsafe / RNA

ENSG00000051341

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 5.9 nTPM (limit 2); bone marrow: 5.9 nTPM (baseline 2)

Cancer RNA. Colon Adenocarcinoma (validation): 398/486 (81.9%); Glioblastoma Multiforme (validation): 47/58 (81.0%); Rectum Adenocarcinoma (validation): 151/207 (72.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Low-fidelity DNA polymerase with a putative helicase activity that promotes microhomology-mediated end-joining (MMEJ), an alternative non-homologous end-joining (NHEJ) machinery required to repair double-strand breaks in DNA during mitosis (PubMed:14576298, PubMed:18503084, PubMed:24648516, PubMed:25642963, PubMed:25643323, PubMed:25775267, PubMed:26636256, PubMed:27311885, PubMed:27591252, PubMed:30655289, PubMed:31562312, ... [full text in table]

Curated tissue note. Highly expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75417 | HPA portal (live; not the pinned values)

POLR2M | Unsafe / RNA

ENSG00000255529

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 18.7 nTPM (limit 10); adrenal gland: 19.4 nTPM (limit 2); amygdala: 20.2 nTPM (limit 2); basal ganglia: 18.1 nTPM (limit 2); bone marrow: 16.7 nTPM (limit 2); cerebellum: 11.1 nTPM (limit 2); cerebral cortex: 18.7 nTPM (limit 2); choroid plexus: 9.5 nTPM (limit 2); colon: 15 nTPM ... [full text in table]

Cancer RNA. Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Appears to be a stable component of the Pol II(G) complex form of RNA polymerase II (Pol II). Pol II synthesizes mRNA precursors and many functional non-coding RNAs and is the central component of the basal RNA polymerase II transcription machinery. May play a role in the Mediator complex-dependent regulation of transcription activation. Acts as a negative regulator of transcriptional activation; this repression is relieved by the ... [full text in table]

Curated tissue note. Detected in adult an fetal brain. Detected in heart, kidney, skeletal muscle, small intestine, lung, prostate and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P0CAP2 | UniProt Q6EEV4 | HPA portal (live; not the pinned values)

POTEG | Unsafe / High RNA

ENSG00000187537

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 47.1 nTPM; blood/total PBMC; hpa_immune_cell_samples; vital; 22.36 TPM; blood/eosinophil; hpa_immune_cell_samples; vital

Cancer RNA. Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6S5H5 | HPA portal (live; not the pinned values)

POU4F1 | Unsafe / Protein

ENSG00000152192

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / inner nuclear layer: Medium (Enhanced); retina / inner plexiform layer: High (Enhanced); retina / nerve fiber layer: High (Enhanced); retina / outer plexiform layer: High (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 19/58 (32.8%); Glioblastoma Multiforme (TCGA): 9/141 (6.4%); Kidney Renal Clear Cell Carcinoma (validation): 4/100 (4.0%)

Cancer IHC. urothelial cancer: 1/10 (10.0%); breast cancer: 0/11 (0.0%)

Biology. Multifunctional transcription factor with different regions mediating its different effects. Acts by binding (via its C-terminal domain) to sequences related to the consensus octamer motif 5'-ATGCAAAT-3' in the regulatory regions of its target genes. Regulates the expression of specific genes involved in differentiation and survival within a subset of neuronal lineages. It has been shown that activation of some of these genes requires ... [full text in table]

Curated tissue note. Expressed in the brain and the retina. Present in the developing brain, spinal cord and eye

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q01851 | HPA portal (live; not the pinned values)

POU5F2 | Unsafe / Protein

ENSG00000248483

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / cells in tubules: Low (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 21/58 (36.2%); Pancreatic Adenocarcinoma (validation): 6/80 (7.5%); Colon Adenocarcinoma (validation): 8/486 (1.6%)

Cancer IHC. testis cancer: 5/12 (41.7%); urothelial cancer: 1/11 (9.1%)

Biology. Transcription factor that binds preferentially to the octamer motif (5'-ATGTTAAT-3'). May exert a regulatory function in meiotic events that are required for terminal differentiation of male germ cell (By similarity)

Curated tissue note. Expressed in skeletal and cardiac muscles, brain, heart and lung. Little or no detectable expression found in pancreas, kidney, liver or placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N7G0 | HPA portal (live; not the pinned values)

PPAN | Unsafe / Protein

ENSG00000130810

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: High (Approved); ... [full text in table]

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Ovary Serous Cystadenocarcinoma (TCGA): 349/349 (100.0%)

Cancer IHC. testis cancer: 11/11 (100.0%); ovarian cancer: 10/11 (90.9%)

Biology. May have a role in cell growth

Curated tissue note. Widely expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NQ55 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PPAT | Unsafe / Protein

ENSG00000128059

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); caudate / glial cells: Medium (Approved); cerebral cortex / endothelial cells: Medium (Approved); cerebral cortex / glial cells: Medium (Approved); colon / glandular cells: Medium (Approved); duodenum / ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. colorectal cancer: 9/11 (81.8%); testis cancer: 7/12 (58.3%)

Biology. Catalyzes the formation of phosphoribosylamine from phosphoribosylpyrophosphate (PRPP) and glutamine

Curated tissue note. Ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q06203 | HPA portal (live; not the pinned values)

PPEF1 | Unsafe / RNA

ENSG00000086717

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 3.5 nTPM (limit 2); spinal cord: 10.3 nTPM (limit 2); cerebral cortex: 3.5 nTPM (baseline 2); spinal cord: 10.3 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 30/50 (60.0%); Glioblastoma Multiforme (validation): 8/58 (13.8%); Skin Cuteneous Melanoma (TCGA): 4/99 (4.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Dephosphorylates and destabilises PDCD5 to inhibit TP53/p53-mediated signaling (PubMed:28051100). May have a role in the recovery or adaptation response of photoreceptors (By similarity)

Curated tissue note. Detected in retina and retinal derived Y-79 retinoblastoma cells (PubMed:9430683). Also found in fetal brain (PubMed:9430683)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14829 | HPA portal (live; not the pinned values)

PPEF2 | Unsafe / RNA

ENSG00000156194

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 231.4 nTPM (limit 2); retina: 231.4 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 15/176 (8.5%); Testicular Germ Cell Tumor (TCGA): 9/133 (6.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 18/282 (6.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May play a role in phototransduction. May dephosphorylate photoactivated rhodopsin. May function as a calcium sensing regulator of ionic currents, energy production or synaptic transmission

Curated tissue note. Retinal specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14830 | HPA portal (live; not the pinned values)

PPM1E | Unsafe / Protein

ENSG00000175175

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / cells in molecular layer: High (Approved); cerebellum / Purkinje cells: High (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Glioblastoma Multiforme (TCGA): 43/141 (30.5%); Prostate Adenocarcinoma (TCGA): 134/480 (27.9%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Protein phosphatase that inactivates multifunctional CaM kinases such as CAMK4 and CAMK2 (By similarity). Dephosphorylates and inactivates PAK. May play a role in the inhibition of actin fiber stress breakdown and in morphological changes driven by TNK2/CDC42. Dephosphorylates PRKAA2 (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WY54 | HPA portal (live; not the pinned values)

PPM1J | Unsafe / Protein

ENSG00000155367

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / cells in molecular layer: Medium (Uncertain); cerebellum / Purkinje ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 48/50 (96.0%); Breast Invasive Carcinoma (TCGA): 679/1022 (66.4%); Ovary Serous Cystadenocarcinoma (validation): 34/81 (42.0%)

Cancer IHC. breast cancer: 6/11 (54.5%); prostate cancer: 5/10 (50.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JR12 | HPA portal (live; not the pinned values)

PPP1R2C | Unsafe / High RNA

ENSG00000102055

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 222.4 nTPM; breast/t-cells; hpa_single_cell_tissue; vital; 86.55 TPM; blood/TREG_MEM; dice_treg_mem; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 17/133 (12.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Functions as a protein phosphatase inhibitor. It inhibits activity of the catalytic subunit of PP1 and weakly inhibits the activity of myosin-associated phosphates

Curated tissue note. Detected in sperm (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14990 | HPA portal (live; not the pinned values)

PPP1R36 | Unsafe / Protein

ENSG00000165807

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): Low (Enhanced); colon / enterocytes - Microvilli: High (Enhanced); duodenum / enterocytes - Microvilli: Low (Enhanced); small intestine / ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 34/80 (42.5%); Pancreatic Adenocarcinoma (TCGA): 55/176 (31.2%); Colon Adenocarcinoma (TCGA): 52/254 (20.5%)

Cancer IHC. testis cancer: 4/12 (33.3%); breast cancer: 0/12 (0.0%)

Biology. Inhibits phosphatase activity of protein phosphatase 1 (PP1) complexes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96LQ0 | HPA portal (live; not the pinned values)

PPP1R42 | Unsafe / High RNA

ENSG00000178125

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 107.7 nTPM; lung/ciliated cells; hpa_single_cell_tissue; vital; 0.7561 TPM; blood/NK; dice_nk; vital

Cancer RNA. Glioblastoma Multiforme (validation): 12/58 (20.7%); Ovary Serous Cystadenocarcinoma (validation): 2/81 (2.5%); Uterine Corpus Endometrial Carcinoma (TCGA): 4/176 (2.3%)

Cancer IHC. liver cancer: 2/12 (16.7%); endometrial cancer: 1/11 (9.1%)

Biology. Regulates phosphatase activity of protein phosphatase 1 (PP1) complexes in the testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z4L9 | HPA portal (live; not the pinned values)

PRAMEF9 | Unsafe / RNA

ENSG00000204505

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skeletal muscle: 32.1 nTPM (limit 2); skeletal muscle: 32.1 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 5/176 (2.8%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

HPA portal (live; not the pinned values)

PRAP1 | Unsafe / Protein

ENSG00000165828

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. duodenum / glandular cells: Medium (Supported); kidney / cells in tubules: Medium (Supported); liver / hepatocytes: Medium (Supported); small intestine / endocrine cells: High (Supported); small intestine / enterocytes: Medium (Supported)

Cancer RNA. Rectum Adenocarcinoma (TCGA): 87/88 (98.9%); Rectum Adenocarcinoma (validation): 203/207 (98.1%); Liver Hepatocellular Carcinoma (validation): 225/231 (97.4%)

Cancer IHC. colorectal cancer: 6/12 (50.0%); liver cancer: 5/12 (41.7%)

Biology. Lipid-binding protein which promotes lipid absorption by facilitating MTTP-mediated lipid transfer (mainly triglycerides and phospholipids) and MTTP-mediated apoB lipoprotein assembly and secretion (By similarity). Protects the gastrointestinal epithelium from irradiation-induced apoptosis (By similarity). May play an important role in maintaining normal growth homeostasis in epithelial cells (PubMed:14583459). Involved in ... [full text in table]

Curated tissue note. Highly expressed in the intestinal epithelial cells (at protein level) (PubMed:32629119). Abundantly expressed in the epithelial cells of the liver, kidney and cervix. Significantly down-regulated in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96NZ9 | HPA portal (live; not the pinned values)

PRH1 | Unsafe / RNA

ENSG00000231887

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. esophagus: 10.7 nTPM (limit 6); pituitary gland: 3.8 nTPM (limit 2); salivary gland: 229.6 nTPM (limit 6); esophagus: 10.7 nTPM (baseline 6); pituitary gland: 3.8 nTPM (baseline 2); salivary gland: 229.6 nTPM (baseline 6)

Cancer RNA. Breast Invasive Carcinoma (validation): 38/50 (76.0%); Skin Cuteneous Melanoma (TCGA): 2/99 (2.0%); Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

HPA portal (live; not the pinned values)

PRKG2 | Unsafe / Protein

ENSG00000138669

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 38/99 (38.4%); Glioblastoma Multiforme (validation): 15/58 (25.9%); Prostate Adenocarcinoma (TCGA): 87/480 (18.1%)

Cancer IHC. liver cancer: 12/12 (100.0%); pancreatic cancer: 10/10 (100.0%)

Biology. Crucial regulator of intestinal secretion and bone growth. Phosphorylates and activates CFTR on the plasma membrane. Plays a key role in intestinal secretion by regulating cGMP-dependent translocation of CFTR in jejunum (PubMed:33106379). Acts downstream of NMDAR to activate the plasma membrane accumulation of GRIA1/GLUR1 in synapse and increase synaptic plasticity. Phosphorylates GRIA1/GLUR1 at Ser-863 (By similarity). Acts as a ... [full text in table]

Curated tissue note. Highly concentrated in brain, lung and intestinal mucosa

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13237 | HPA portal (live; not the pinned values)

PRM1 | Unsafe / High RNA

ENSG00000175646

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 1360 nTPM; testis/macrophages; hpa_single_cell_tissue; vital; 2.17 TPM; blood/TREG_MEM; dice_treg_mem; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 19/133 (14.3%); Glioblastoma Multiforme (TCGA): 1/141 (0.7%); Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%)

Cancer IHC. colorectal cancer: 4/12 (33.3%); skin cancer: 2/12 (16.7%)

Biology. Protamines substitute for histones in the chromatin of sperm during the haploid phase of spermatogenesis. They compact sperm DNA into a highly condensed, stable and inactive complex

Curated tissue note. Testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P04553 | HPA portal (live; not the pinned values)

PRM2 | Unsafe / High RNA

ENSG00000122304

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 1074 nTPM; testis/macrophages; hpa_single_cell_tissue; vital; 1.052 TPM; blood/TH17; dice_th17; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 19/133 (14.3%); Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Protamines substitute for histones in the chromatin of sperm during the haploid phase of spermatogenesis. They compact sperm DNA into a highly condensed, stable and inactive complex

Curated tissue note. Testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P04554 | HPA portal (live; not the pinned values)

PRM3 | Unsafe / RNA

ENSG00000178257

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.6 nTPM (limit 2); bone marrow: 2.6 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 1/282 (0.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Protamines substitute for histones in the chromatin of sperm during the haploid phase of spermatogenesis. They compact sperm DNA into a highly condensed, stable and inactive complex (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NNZ6 | HPA portal (live; not the pinned values)

PRND | Unsafe / RNA

ENSG00000171864

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 39.4 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 35/133 (26.3%); Breast Invasive Carcinoma (validation): 11/50 (22.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 60/521 (11.5%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Required for normal acrosome reaction and for normal male fertility (By similarity). Can bind Cu(2+) (PubMed:15218028, PubMed:20411530)

Curated tissue note. Expressed in testis, in Sertoli cells, ejaculated spermatozoa and in seminal fluid (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UKY0 | HPA portal (live; not the pinned values)

PROCA1 | Unsafe / RNA

ENSG00000167525

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 4 nTPM (limit 2); basal ganglia: 5.1 nTPM (limit 2); cerebellum: 7.2 nTPM (limit 4); cerebral cortex: 4.6 nTPM (limit 4); choroid plexus: 4.8 nTPM (limit 2); hippocampal formation: 4 nTPM (limit 2); hypothalamus: 4.9 nTPM (limit 2); kidney: 8 nTPM (limit 4); midbrain: 4.3 nTPM (limit 2); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 41/50 (82.0%); Testicular Germ Cell Tumor (TCGA): 93/133 (69.9%); Kidney Renal Papillary Cell Carcinoma (TCGA): 80/282 (28.4%)

Cancer IHC. liver cancer: 1/12 (8.3%); renal cancer: 1/12 (8.3%)

Biology. High expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCQ7 | HPA portal (live; not the pinned values)

PROK2 | Unsafe / RNA

ENSG00000163421

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 63.7 nTPM (limit 4); lung: 12.7 nTPM (limit 4); spleen: 21.9 nTPM (limit 20); bone marrow: 63.7 nTPM (baseline 2); liver: 2.4 nTPM (baseline 2); lung: 12.7 nTPM (baseline 2); spleen: 21.9 nTPM (baseline 10)

Cancer RNA. Colon Adenocarcinoma (validation): 91/486 (18.7%); Rectum Adenocarcinoma (validation): 32/207 (15.5%); Ovary Serous Cystadenocarcinoma (TCGA): 35/349 (10.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May function as an output molecule from the suprachiasmatic nucleus (SCN) that transmits behavioral circadian rhythm. May also function locally within the SCN to synchronize output. Potently contracts gastrointestinal (GI) smooth muscle

Curated tissue note. Expressed in the testis and, at low levels, in the small intestine

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HC23 | HPA portal (live; not the pinned values)

PRORP | Unsafe / Protein

ENSG00000100890

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: High (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / ciliated cells (tip of cilia): Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); ovarian cancer: 12/12 (100.0%)

Biology. Catalytic ribonuclease component of mitochondrial ribonuclease P, a complex composed of TRMT10C/MRPP1, HSD17B10/MRPP2 and PRORP/MRPP3, which cleaves tRNA molecules in their 5'-ends (PubMed:18984158, PubMed:25953853, PubMed:34715011). The presence of TRMT10C/MRPP1, HSD17B10/MRPP2 is required to catalyze tRNA molecules in their 5'-ends (PubMed:25953853)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O15091 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PRPH2 | Unsafe / Protein

ENSG00000112619

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: High (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 9/50 (18.0%); Kidney Chromophobe (TCGA): 3/64 (4.7%); Thyroid Carcinoma (TCGA): 21/495 (4.2%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Essential for retina photoreceptor outer segment disk morphogenesis, may also play a role with ROM1 in the maintenance of outer segment disk structure (By similarity). Required for the maintenance of retinal outer nuclear layer thickness (By similarity). Required for the correct development and organization of the photoreceptor inner segment (By similarity)

Curated tissue note. Retina (photoreceptor). In rim region of ROS (rod outer segment) disks

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P23942 | HPA portal (live; not the pinned values)

PRR11 | Unsafe / RNA

ENSG00000068489

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 17.8 nTPM (limit 2); duodenum: 6.3 nTPM (limit 2); lung: 2.2 nTPM (limit 2); lymph node: 10 nTPM (limit 6); small intestine: 4.8 nTPM (limit 2); smooth muscle: 3.5 nTPM (limit 2); thymus: 20 nTPM (limit 10); bone marrow: 17.8 nTPM (baseline 2); duodenum: 6.3 nTPM (baseline 2); lung: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 204/207 (98.6%)

Cancer IHC. cervical cancer: 12/12 (100.0%); skin cancer: 11/11 (100.0%)

Biology. Plays a critical role in cell cycle progression

Curated tissue note. Ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96HE9 | HPA portal (live; not the pinned values)

PRR19 | Unsafe / RNA

ENSG00000188368

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.1 nTPM (limit 2); basal ganglia: 4.1 nTPM (limit 2); bone marrow: 2.2 nTPM (limit 2); cerebellum: 2.2 nTPM (limit 2); cerebral cortex: 2.3 nTPM (limit 2); amygdala: 2.1 nTPM (baseline 2); basal ganglia: 4.1 nTPM (baseline 2); bone marrow: 2.2 nTPM (baseline 2); cerebellum: 2.2 nTPM ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 42/50 (84.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 129/176 (73.3%); Testicular Germ Cell Tumor (TCGA): 83/133 (62.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Promotes meiotic crossing over formation through its interaction with CNTD1 by participating in the crossover differentiation step of crossover-specific recombination intermediates

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NJB7 | HPA portal (live; not the pinned values)

PRR22 | Unsafe / RNA

ENSG00000212123

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.1 nTPM (limit 2); basal ganglia: 4.2 nTPM (limit 2); hippocampal formation: 3.5 nTPM (limit 2); hypothalamus: 2.7 nTPM (limit 2); midbrain: 2.5 nTPM (limit 2); pancreas: 6.1 nTPM (limit 2); pituitary gland: 3.7 nTPM (limit 2); spinal cord: 2.8 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Testicular Germ Cell Tumor (TCGA): 108/133 (81.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 78/176 (44.3%)

Cancer IHC. glioma: 1/11 (9.1%); breast cancer: 0/9 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZ63 | HPA portal (live; not the pinned values)

PRR27 | Unsafe / Protein

ENSG00000187533

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. salivary gland / glandular cells: High (Enhanced)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 3/176 (1.7%); Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%); Breast Invasive Carcinoma (TCGA): 13/1022 (1.3%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6MZM9 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PRSS21 | Unsafe / Protein

ENSG00000007038

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. lung / macrophages: Medium (Enhanced)

Cancer RNA. Ovary Serous Cystadenocarcinoma (validation): 69/81 (85.2%); Ovary Serous Cystadenocarcinoma (TCGA): 270/349 (77.4%); Lung Squamous Cell Carcinoma (TCGA): 276/489 (56.4%)

Cancer IHC. breast cancer: 1/12 (8.3%); cervical cancer: 0/12 (0.0%)

Biology. Could regulate proteolytic events associated with testicular germ cell maturation

Curated tissue note. Expressed predominantly in premeiotic testicular germ cells, mostly late pachytene and diplotene spermatocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y6M0 | HPA portal (live; not the pinned values)

PSEN1 | Unsafe / Protein

ENSG00000080815

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / basal cells: High (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / goblet cells: High (Approved); caudate / glial cells: Low (Approved); caudate / ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. prostate cancer: 10/12 (83.3%); colorectal cancer: 8/12 (66.7%)

Biology. Catalytic subunit of the gamma-secretase complex, an endoprotease complex that catalyzes the intramembrane cleavage of integral membrane proteins such as Notch receptors and APP (amyloid-beta precursor protein) (PubMed:10206644, PubMed:10545183, PubMed:10593990, PubMed:10811883, PubMed:10899933, PubMed:12679784, PubMed:12740439, PubMed:15274632, PubMed:20460383, PubMed:25043039, PubMed:26280335, PubMed:28269784, PubMed:30598546, ... [full text in table]

Curated tissue note. Detected in azurophile granules in neutrophils and in platelet cytoplasmic granules (at protein level) (PubMed:11987239). Expressed in a wide range of tissues including various regions of the brain, liver, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P49768 | HPA portal (live; not the pinned values)

PSG4 | Unsafe / RNA

ENSG00000243137

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. parathyroid gland: 53.8 nTPM (limit 12)

Cancer RNA. Kidney Chromophobe (TCGA): 26/64 (40.6%); Bladder Urothelial Carcinoma (TCGA): 17/169 (10.1%); Head and Neck Squamous Cell Carcinoma (TCGA): 37/492 (7.5%)

Cancer IHC. liver cancer: 1/11 (9.1%); melanoma: 1/12 (8.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q00888 | HPA portal (live; not the pinned values)

PSG5 | Unsafe / High RNA

ENSG00000204941

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 31.6 nTPM; placenta/mixed immune cells; hpa_single_cell_tissue; vital; 0.7 TPM; blood/NK-cell; hpa_immune_monaco; vital

Cancer RNA. Kidney Chromophobe (TCGA): 30/64 (46.9%); Lung Squamous Cell Carcinoma (validation): 5/68 (7.4%); Bladder Urothelial Carcinoma (TCGA): 9/169 (5.3%)

Cancer IHC. liver cancer: 1/11 (9.1%); melanoma: 1/12 (8.3%)

Biology. Synthesized by syncytiotrophoblast of the placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15238 | HPA portal (live; not the pinned values)

PSKH2 | Unsafe / Protein

ENSG00000147613

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / endothelial cells: Medium (Approved); colon / endothelial cells: Medium (Approved); kidney / cells in glomeruli: Medium (Approved); lung / macrophages: High (Approved); oral mucosa / squamous epithelial cells: Medium (Approved); skin 1 / fibroblasts: Medium (Approved); skin 2 / ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 13/133 (9.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 1/283 (0.4%)

Cancer IHC. melanoma: 3/10 (30.0%); endometrial cancer: 2/12 (16.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96QS6 | HPA portal (live; not the pinned values)

PSMA2 | Unsafe / Protein

ENSG00000106588

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. prostate cancer: 9/11 (81.8%); renal cancer: 9/12 (75.0%)

Biology. Component of the 20S core proteasome complex involved in the proteolytic degradation of most intracellular proteins. This complex plays numerous essential roles within the cell by associating with different regulatory particles. Associated with two 19S regulatory particles, forms the 26S proteasome and thus participates in the ATP-dependent degradation of ubiquitinated proteins. The 26S proteasome plays a key role in the maintenance of ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P25787 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PSMB11 | Unsafe / Protein

ENSG00000222028

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. thymus / cortical cells: Medium (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. The proteasome is a multicatalytic proteinase complex which is characterized by its ability to cleave peptides with Arg, Phe, Tyr, Leu, and Glu adjacent to the leaving group at neutral or slightly basic pH. The proteasome has an ATP-dependent proteolytic activity. Incorporated instead of PSMB5 or PSMB8, this unit reduces the chymotrypsin-like activity of the proteasome (By similarity). Plays a pivotal role in development of ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A5LHX3 | HPA portal (live; not the pinned values)

PSORS1C1 | Unsafe / High RNA

ENSG00000204540

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 44.6 nTPM; colon/distal enterocytes; hpa_single_cell_tissue; protected; 0.2388 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 32/50 (64.0%); Pancreatic Adenocarcinoma (validation): 31/80 (38.8%); Ovary Serous Cystadenocarcinoma (validation): 31/81 (38.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Expressed in skin. Also found in heart, placenta, liver, skeletal muscle and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UIG5 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

PSPC1 | Unsafe / Protein

ENSG00000121390

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Supported); adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: Medium (Supported); caudate / glial cells: High (Supported); caudate / neuronal cells: High (Supported); cerebellum / cells in granular layer: High (Supported); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. RNA-binding protein required for the formation of nuclear paraspeckles (PubMed:22416126). Binds to poly(A), poly(G) and poly(U) RNA homopolymers (PubMed:22416126). Regulates, cooperatively with NONO and SFPQ, androgen receptor-mediated gene transcription activity in Sertoli cell line (By similarity). Regulates the circadian clock by repressing the transcriptional activator activity of the CLOCK-BMAL1 heterodimer (By similarity). Plays ... [full text in table]

Curated tissue note. Expressed in pancreas, kidney, skeletal muscle, liver, lung, placenta, brain and heart

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WXF1 | HPA portal (live; not the pinned values)

PSPH | Unsafe / Protein

ENSG00000146733

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / basal cells: Low (Approved); bronchus / ciliated cells (cell body): Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Medium (Approved); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); ovarian cancer: 11/12 (91.7%)

Biology. Catalyzes the last irreversible step in the biosynthesis of L-serine from carbohydrates, the dephosphorylation of O-phospho-L-serine to L-serine (PubMed:12213811, PubMed:14673469, PubMed:15291819, PubMed:25080166, PubMed:9222972). L-serine can then be used in protein synthesis, to produce other amino acids, in nucleotide metabolism or in glutathione synthesis, or can be racemized to D-serine, a neuromodulator (PubMed:14673469). May ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P78330 | HPA portal (live; not the pinned values)

PSTK | Unsafe / Protein

ENSG00000179988

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 44/50 (88.0%); Ovary Serous Cystadenocarcinoma (validation): 40/81 (49.4%); Testicular Germ Cell Tumor (TCGA): 61/133 (45.9%)

Cancer IHC. ovarian cancer: 12/12 (100.0%); liver cancer: 11/11 (100.0%)

Biology. Specifically phosphorylates seryl-tRNA(Sec) to O-phosphoseryl-tRNA(Sec), an activated intermediate for selenocysteine biosynthesis. No activity with other tRNAs has been detected

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IV42 | HPA portal (live; not the pinned values)

PTBP2 | Unsafe / Protein

ENSG00000117569

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. esophagus / squamous epithelial cells: Low (Uncertain); pancreas / exocrine glandular cells: Medium (Uncertain)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 134/141 (95.0%); Pancreatic Adenocarcinoma (validation): 76/80 (95.0%)

Cancer IHC. renal cancer: 5/12 (41.7%); liver cancer: 1/10 (10.0%)

Biology. RNA-binding protein which binds to intronic polypyrimidine tracts and mediates negative regulation of exons splicing. May antagonize in a tissue-specific manner the ability of NOVA1 to activate exon selection. In addition to its function in pre-mRNA splicing, plays also a role in the regulation of translation Reduced affinity for RNA

Curated tissue note. Mainly expressed in brain although also detected in other tissues like heart and skeletal muscle. Isoform 1 and isoform 2 are specifically expressed in neuronal tissues. Isoform 3 and isoform 4 are expressed ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UKA9 | HPA portal (live; not the pinned values)

PTCH1 | Unsafe / Protein

ENSG00000185920

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: High (Uncertain); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Colon Adenocarcinoma (validation): 484/486 (99.6%); Glioblastoma Multiforme (validation): 54/58 (93.1%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Negative regulator of the smoothened signaling pathway, which acts as a receptor for hedgehog (SHH, IHH and DHH) morphogens (PubMed:21537345, PubMed:29954986, PubMed:29995851, PubMed:30139912, PubMed:31127104, PubMed:31555730, PubMed:37611095, PubMed:40128518, PubMed:9811851). In absence of hedgehog, acts as an inhibitor of smoothened protein (SMO) by preventing SMO access to cholesterol: mechanistically, acts as a cholesterol ... [full text in table]

Curated tissue note. In the adult, expressed in brain, lung, liver, heart, placenta, skeletal muscle, pancreas and kidney. Expressed in tumor cells but not in normal skin

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13635 | HPA portal (live; not the pinned values)

PTF1A | Unsafe / RNA

ENSG00000168267

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pancreas: 101.2 nTPM (limit 2); pancreas: 101.2 nTPM (baseline 2)

Cancer RNA. Pancreatic Adenocarcinoma (TCGA): 29/176 (16.5%); Rectum Adenocarcinoma (TCGA): 5/88 (5.7%); Rectum Adenocarcinoma (validation): 7/207 (3.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcription factor implicated in the cell fate determination in various organs. Binds to the E-box consensus sequence 5'-CANNTG-3'. Plays a role in early and late pancreas development and differentiation. Important for determining whether cells allocated to the pancreatic buds continue towards pancreatic organogenesis or revert back to duodenal fates. May be involved in the maintenance of exocrine pancreas-specific gene expression ... [full text in table]

Curated tissue note. Pancreas-specific (at protein level). Loss of expression is seen in ductal type pancreas cancers

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7RTS3 | HPA portal (live; not the pinned values)

PTGR2 | Unsafe / Protein

ENSG00000140043

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular layer: Medium ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 229/231 (99.1%); Kidney Chromophobe (TCGA): 63/64 (98.4%); Prostate Adenocarcinoma (TCGA): 472/480 (98.3%)

Cancer IHC. prostate cancer: 11/11 (100.0%); breast cancer: 6/9 (66.7%)

Biology. Functions as 15-oxo-prostaglandin 13-reductase and acts on 15-keto-PGE1, 15-keto-PGE2, 15-keto-PGE1-alpha and 15-keto-PGE2-alpha with highest activity towards 15-keto-PGE2 (PubMed:19000823). Overexpression represses transcriptional activity of PPARG and inhibits adipocyte differentiation (By similarity)

Curated tissue note. Widely expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N8N7 | HPA portal (live; not the pinned values)

PTPN11 | Unsafe / Protein

ENSG00000179295

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Approved); adrenal gland / glandular cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: High (Approved); cerebellum / cells in granular layer: High (Approved); cerebellum / cells in molecular layer: High ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Colon Adenocarcinoma (validation): 486/486 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%)

Cancer IHC. prostate cancer: 11/11 (100.0%); urothelial cancer: 11/11 (100.0%)

Biology. Acts downstream of various receptor and cytoplasmic protein tyrosine kinases to participate in the signal transduction from the cell surface to the nucleus (PubMed:10655584, PubMed:14739280, PubMed:18559669, PubMed:18829466, PubMed:26742426, PubMed:28074573, PubMed:32184441). Positively regulates MAPK signal transduction pathway (PubMed:28074573). Dephosphorylates GAB1, ARHGAP35 and EGFR (PubMed:28074573). Dephosphorylates ROCK2 at ... [full text in table]

Curated tissue note. Widely expressed, with highest levels in heart, brain, and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q06124 | HPA portal (live; not the pinned values)

PTPRQ | Unsafe / RNA

ENSG00000139304

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 6.9 nTPM (limit 2); lung: 2.2 nTPM (limit 2); kidney: 6.9 nTPM (baseline 2); lung: 2.2 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 3/50 (6.0%); Bladder Urothelial Carcinoma (TCGA): 8/169 (4.7%); Lung Squamous Cell Carcinoma (validation): 2/68 (2.9%)

Cancer IHC. breast cancer: 9/12 (75.0%); melanoma: 7/10 (70.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

HPA portal (live; not the pinned values)

PTRH1 | Unsafe / Protein

ENSG00000187024

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Ovary Serous Cystadenocarcinoma (TCGA): 349/349 (100.0%)

Cancer IHC. colorectal cancer: 10/12 (83.3%); prostate cancer: 5/10 (50.0%)

Biology. Peptidyl-tRNA hydrolase that cleaves nascent chains-tRNAs that are not stably fixed in the P-site of 60S ribosome-nascent chain complexes (PubMed:30244831). Acts downstream of the ribosome-associated quality control (RQC) pathway to release non-ubiquitinated nascent chains from 60S and 80S ribosome-nascent chain complexes (PubMed:30244831). Does not act on ubiquitinated nascent chains, which are cleaved by ANKZF1 for degradation ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86Y79 | HPA portal (live; not the pinned values)

PTTG1 | Unsafe / Protein

ENSG00000164611

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. 608.3 nTPM; liver/erythroid cells; hpa_single_cell_tissue; vital; 238.2 TPM; blood/T-reg; hpa_immune_cell_samples; vital

Cancer RNA. Thyroid Carcinoma (TCGA): 495/495 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%)

Cancer IHC. endometrial cancer: 7/12 (58.3%); liver cancer: 6/11 (54.5%)

Biology. Regulatory protein, which plays a central role in chromosome stability, in the p53/TP53 pathway, and DNA repair. Probably acts by blocking the action of key proteins. During the mitosis, it blocks Separase/ESPL1 function, preventing the proteolysis of the cohesin complex and the subsequent segregation of the chromosomes. At the onset of anaphase, it is ubiquitinated, conducting to its destruction and to the liberation of ESPL1. Its ... [full text in table]

Curated tissue note. Expressed at low level in most tissues, except in adult testis, where it is highly expressed. Overexpressed in many patients suffering from pituitary adenomas, primary epithelial neoplasias, and esophageal ... [full text in table]

Therapeutic evidence. Trial registration only.

UniProt O95997 | Trial/source record | HPA portal (live; not the pinned values)

PURG | Unsafe / Protein

ENSG00000172733

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / molecular layer cells - nucleus: Low (Enhanced); cerebral cortex / glial cells: Low (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); colon / peripheral nerve/ganglion: Low (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Glioblastoma Multiforme (TCGA): 65/141 (46.1%); Kidney Chromophobe (TCGA): 8/64 (12.5%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Isoform 1 is expressed in testis and glioblastoma. Isoform 2 is expressed in fetal lung

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UJV8 | HPA portal (live; not the pinned values)

QRICH2 | Unsafe / RNA

ENSG00000129646

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.2 nTPM (limit 2); basal ganglia: 3.2 nTPM (limit 2); hippocampal formation: 2.5 nTPM (limit 2); hypothalamus: 3.1 nTPM (limit 2); midbrain: 2.3 nTPM (limit 2); retina: 2.6 nTPM (limit 2); spinal cord: 2.4 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 38/50 (76.0%); Glioblastoma Multiforme (validation): 41/58 (70.7%); Testicular Germ Cell Tumor (TCGA): 50/133 (37.6%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Has an essential role in the formation of sperm flagella and flagellar structure maintainance. It acts as a suppressor of ubiquitination and degradation of proteins involved in flagellar development and motility

Curated tissue note. Expressed in the sperm

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0J4 | HPA portal (live; not the pinned values)

R3HCC1L | Unsafe / Protein

ENSG00000166024

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in granular layer: Low (Enhanced); cerebellum / cells in molecular layer: Low ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. melanoma: 12/12 (100.0%); colorectal cancer: 11/11 (100.0%)

Biology. Expressed in placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z5L2 | HPA portal (live; not the pinned values)

R3HDML | Unsafe / Protein

ENSG00000101074

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / goblet cells: Low (Enhanced); duodenum / goblet cells: Low (Enhanced); kidney / collecting ducts: Medium (Enhanced); kidney / distal tubules: Medium (Enhanced); small intestine / goblet cells: Medium (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (validation): 197/207 (95.2%); Colon Adenocarcinoma (validation): 315/486 (64.8%); Rectum Adenocarcinoma (TCGA): 55/88 (62.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Putative serine protease inhibitor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H3Y0 | HPA portal (live; not the pinned values)

RAB11B | Unsafe / RNA

ENSG00000185236

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 98.8 nTPM (limit 10); adrenal gland: 93.7 nTPM (limit 2); amygdala: 123.8 nTPM (limit 2); basal ganglia: 137.8 nTPM (limit 2); bone marrow: 67.7 nTPM (limit 2); cerebellum: 138.7 nTPM (limit 2); cerebral cortex: 138.7 nTPM (limit 2); choroid plexus: 64.4 nTPM (limit 2); colon: 95.8 ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. The small GTPases Rab are key regulators of intracellular membrane trafficking, from the formation of transport vesicles to their fusion with membranes. Rabs cycle between an inactive GDP-bound form and an active GTP-bound form that is able to recruit to membranes different set of downstream effectors directly responsible for vesicle formation, movement, tethering and fusion (PubMed:14627637, PubMed:19029296, PubMed:19244346, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15907 | HPA portal (live; not the pinned values)

RAB27B | Unsafe / Protein

ENSG00000041353

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); cerebellum / Purkinje cells: Low (Enhanced); cerebral cortex / neuropil: Low (Enhanced); colon / glandular cells: High (Enhanced); duodenum / glandular cells: High (Enhanced); esophagus / ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 76/80 (95.0%); Prostate Adenocarcinoma (TCGA): 453/480 (94.4%); Breast Invasive Carcinoma (validation): 47/50 (94.0%)

Cancer IHC. prostate cancer: 12/12 (100.0%); breast cancer: 10/12 (83.3%)

Biology. The small GTPases Rab are key regulators of intracellular membrane trafficking, from the formation of transport vesicles to their fusion with membranes. Rabs cycle between an inactive GDP-bound form and an active GTP-bound form that is able to recruit to membranes different sets of downstream effectors directly responsible for vesicle formation, movement, tethering and fusion (PubMed:21775604, PubMed:30771381). RAB27B regulates ... [full text in table]

Curated tissue note. Expressed primarily in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00194 | HPA portal (live; not the pinned values)

RAB41 | Unsafe / RNA

ENSG00000147127

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 56 nTPM (limit 2); retina: 56 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (TCGA): 53/141 (37.6%); Kidney Renal Papillary Cell Carcinoma (TCGA): 10/282 (3.5%); Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%)

Cancer IHC. pancreatic cancer: 12/12 (100.0%); prostate cancer: 11/11 (100.0%)

Biology. The small GTPases Rab are key regulators of intracellular membrane trafficking, from the formation of transport vesicles to their fusion with membranes. Rabs cycle between an inactive GDP-bound form and an active GTP-bound form that is able to recruit to membranes different sets of downstream effectors directly responsible for vesicle formation, movement, tethering and fusion (PubMed:23936529). RAB41 is required for normal Golgi ribbon ... [full text in table]

Curated tissue note. Widely expressed in brain, testis, lung, heart, ovary, colon, kidney, uterus and spleen but not in liver

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JT25 | HPA portal (live; not the pinned values)

RABGEF1 | Unsafe / RNA

ENSG00000154710

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 15.7 nTPM (limit 10); adrenal gland: 13.8 nTPM (limit 2); amygdala: 5.4 nTPM (limit 2); basal ganglia: 6.3 nTPM (limit 2); bone marrow: 76 nTPM (limit 2); cerebellum: 13 nTPM (limit 2); cerebral cortex: 12.5 nTPM (limit 2); choroid plexus: 13.5 nTPM (limit 2); colon: 10.5 nTPM ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Acts as a guanine-nucleotide releasing factor (GEF) for RAB5A by promoting the conversion of inactive RAB-GDP to the active form RAB-GTP (PubMed:15339665, PubMed:9323142). Forms a complex with RABEP1 that acts as Rab4/Rab5 effector regulating endosome fusion. Involved in endocytic membrane fusion and membrane trafficking of recycling endosomes (PubMed:11452015, PubMed:9323142). Also functions as a ubiquitin ligase (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UJ41 | HPA portal (live; not the pinned values)

RABL3 | Unsafe / Protein

ENSG00000144840

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Adenocarcinoma (validation): 102/105 (97.1%)

Cancer IHC. colorectal cancer: 6/12 (50.0%); lung cancer: 6/12 (50.0%)

Biology. Small GTPase required for KRAS signaling regulation and modulation of cell proliferation (PubMed:31406347). Regulator of KRAS prenylation, and probably prenylation of other small GTPases (PubMed:31406347). Required for lymphocyte development and function (By similarity). Not required for myeloid cell development (By similarity). Interacts with Rab11 to promote ciliary vesicle formation at the mother centriole, thereby regulating early ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5HYI8 | HPA portal (live; not the pinned values)

RAD51 | Unsafe / Protein

ENSG00000051180

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); caudate / neuronal cells: Low (Enhanced); colon / glandular cells: Low (Enhanced); duodenum / glandular cells: Low (Enhanced); esophagus / ... [full text in table]

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 273/283 (96.5%); Colon Adenocarcinoma (TCGA): 245/254 (96.5%); Testicular Germ Cell Tumor (TCGA): 128/133 (96.2%)

Cancer IHC. testis cancer: 11/11 (100.0%); skin cancer: 7/9 (77.8%)

Biology. Homologous DNA recombinase that catalyzes strand exchange, a key step in DNA repair through homologous recombination (HR) (PubMed:12205100, PubMed:12442171, PubMed:15937124, PubMed:17515904, PubMed:17515903, PubMed:18417535, PubMed:19303847, PubMed:20231364, PubMed:20348101, PubMed:22325354, PubMed:23509288, PubMed:23754376, PubMed:24141787, PubMed:26681308, PubMed:27941124, PubMed:28575658, PubMed:32640219, PubMed:37499663, ... [full text in table]

Curated tissue note. Highly expressed in testis and thymus, followed by small intestine, placenta, colon, pancreas and ovary (PubMed:18417535). Weakly expressed in breast (PubMed:18417535)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q06609 | HPA portal (live; not the pinned values)

RAD51AP1 | Unsafe / RNA

ENSG00000111247

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 16.7 nTPM (limit 2); duodenum: 5 nTPM (limit 2); lymph node: 8.7 nTPM (limit 6); rectum: 6.3 nTPM (limit 6); small intestine: 4.7 nTPM (limit 2); thymus: 14 nTPM (limit 10); bone marrow: 16.7 nTPM (baseline 2); duodenum: 5 nTPM (baseline 2); lymph node: 8.7 nTPM (baseline 6); rectum: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Squamous Cell Carcinoma (validation): 67/68 (98.5%); Colon Adenocarcinoma (TCGA): 236/254 (92.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Structure-specific DNA-binding protein involved in DNA repair by promoting RAD51-mediated homologous recombination (PubMed:17996710, PubMed:17996711, PubMed:20871616, PubMed:25288561, PubMed:26323318). Acts by stimulating D-Loop formation by RAD51: specifically enhances joint molecule formation through its structure-specific DNA interaction and its interaction with RAD51 (PubMed:17996710, PubMed:17996711). Binds single-stranded DNA ... [full text in table]

Curated tissue note. Highly expressed in testis and thymus (PubMed:9396801). Lower levels in colon and small intestine (PubMed:9396801). Little or no expression in spleen, prostate, ovary and peripheral blood leukocytes ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96B01 | HPA portal (live; not the pinned values)

RAD51B | Unsafe / Protein

ENSG00000182185

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Enhanced); adrenal gland / glandular cells: High (Enhanced); bone marrow / hematopoietic cells: High (Enhanced); bronchus / respiratory epithelial cells: High (Enhanced); caudate / glial cells: Medium (Enhanced); caudate / neuronal cells: High (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Pancreatic Adenocarcinoma (validation): 75/80 (93.8%); Rectum Adenocarcinoma (validation): 187/207 (90.3%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Involved in the homologous recombination repair (HRR) pathway of double-stranded DNA breaks arising during DNA replication or induced by DNA-damaging agents. May promote the assembly of presynaptic RAD51 nucleoprotein filaments. Binds single-stranded DNA and double-stranded DNA and has DNA-dependent ATPase activity. Part of the RAD51 paralog protein complex BCDX2 which acts in the BRCA1-BRCA2-dependent HR pathway. Upon DNA damage, ... [full text in table]

Curated tissue note. Expressed in a wide range of tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O15315 | HPA portal (live; not the pinned values)

RAD54B | Unsafe / Protein

ENSG00000197275

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / Purkinje cells: Low (Enhanced); cerebral cortex / endothelial cells: Low (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 44/50 (88.0%); Colon Adenocarcinoma (validation): 375/486 (77.2%)

Cancer IHC. urothelial cancer: 6/12 (50.0%); lymphoma: 4/12 (33.3%)

Biology. Multifunctional ATPase that could play with RAD54, a redundant role in homologous recombination (HR), a major pathway for repairing DNA double-strand breaks (DSBs), single-stranded DNA (ssDNA) gaps, and stalled or collapsed replication forks (PubMed:11884632, PubMed:16428451). Could act as a molecular motor during the homology search and guide RAD51 ssDNA along a donor dsDNA thereby changing the homology search from the diffusion-based ... [full text in table]

Curated tissue note. Abundantly expressed in testis and spleen. Relatively low levels observed in thymus, prostate, ovary and colon

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y620 | HPA portal (live; not the pinned values)

RAD54L | Unsafe / Protein

ENSG00000085999

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 466/486 (95.9%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 270/283 (95.4%); Testicular Germ Cell Tumor (TCGA): 124/133 (93.2%)

Cancer IHC. colorectal cancer: 9/11 (81.8%); glioma: 9/11 (81.8%)

Biology. Multifunctional ATPase that plays a role in homologous recombination (HR) which is a major pathway for repairing DNA double-strand breaks (DSBs), single-stranded DNA (ssDNA) gaps, and stalled or collapsed replication forks (PubMed:11459989, PubMed:12205100, PubMed:24798879, PubMed:27264870, PubMed:32457312, PubMed:9774452). Acts as a molecular motor during the homology search and guides RAD51 ssDNA along a donor dsDNA thereby changing ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92698 | HPA portal (live; not the pinned values)

RAD9B | Unsafe / Protein

ENSG00000151164

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. skeletal muscle / myocytes: Medium (Approved)

Cancer RNA. Breast Invasive Carcinoma (validation): 36/50 (72.0%); Glioblastoma Multiforme (validation): 8/58 (13.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 15/176 (8.5%)

Cancer IHC. melanoma: 10/12 (83.3%); breast cancer: 8/12 (66.7%)

Biology. Expressed in testis and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6WBX8 | HPA portal (live; not the pinned values)

RAG1 | Unsafe / RNA

ENSG00000166349

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 4.2 nTPM (limit 2); thymus: 385 nTPM (limit 10); bone marrow: 4.2 nTPM (baseline 2); thymus: 385 nTPM (baseline 10)

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 5/68 (7.4%); Glioblastoma Multiforme (validation): 3/58 (5.2%); Pancreatic Adenocarcinoma (validation): 2/80 (2.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Catalytic component of the RAG complex, a multiprotein complex that mediates the DNA cleavage phase during V(D)J recombination. V(D)J recombination assembles a diverse repertoire of immunoglobulin and T-cell receptor genes in developing B and T-lymphocytes through rearrangement of different V (variable), in some cases D (diversity), and J (joining) gene segments. In the RAG complex, RAG1 mediates the DNA-binding to the conserved ... [full text in table]

Curated tissue note. Maturing lymphoid cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P15918 | HPA portal (live; not the pinned values)

RAG2 | Unsafe / RNA

ENSG00000175097

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. thymus: 146.4 nTPM (limit 10); thyroid gland: 23 nTPM (limit 10); thymus: 146.4 nTPM (baseline 10); thyroid gland: 23 nTPM (baseline 10)

Cancer RNA. Thyroid Carcinoma (TCGA): 36/495 (7.3%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Core component of the RAG complex, a multiprotein complex that mediates the DNA cleavage phase during V(D)J recombination. V(D)J recombination assembles a diverse repertoire of immunoglobulin and T-cell receptor genes in developing B and T-lymphocytes through rearrangement of different V (variable), in some cases D (diversity), and J (joining) gene segments. DNA cleavage by the RAG complex occurs in 2 steps: a first nick is introduced ... [full text in table]

Curated tissue note. Cells of the B- and T-lymphocyte lineages

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P55895 | HPA portal (live; not the pinned values)

RALGPS2 | Unsafe / Protein

ENSG00000116191

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); bronchus / ciliated cells (cell body): Low (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); cerebellum / Purkinje cells: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); colon / endothelial cells: Low (Enhanced); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. melanoma: 3/12 (25.0%); glioma: 1/11 (9.1%)

Biology. Guanine nucleotide exchange factor for the small GTPase RALA. May be involved in cytoskeletal organization. May also be involved in the stimulation of transcription in a Ras-independent fashion (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86X27 | HPA portal (live; not the pinned values)

RANBP17 | Unsafe / High RNA

ENSG00000204764

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 382.9 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 3.986 TPM; blood/NK; dice_nk; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 41/50 (82.0%); Pancreatic Adenocarcinoma (validation): 52/80 (65.0%); Glioblastoma Multiforme (validation): 15/58 (25.9%)

Cancer IHC. liver cancer: 8/12 (66.7%); ovarian cancer: 4/11 (36.4%)

Biology. May function as a nuclear transport receptor

Curated tissue note. Highly expressed in testis, moderately in pancreas and weakly in other tissues studied

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H2T7 | HPA portal (live; not the pinned values)

RAPGEF6 | Unsafe / Protein

ENSG00000158987

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. ovarian cancer: 12/12 (100.0%); breast cancer: 11/12 (91.7%)

Biology. Guanine nucleotide exchange factor (GEF) for Rap1A, Rap2A and M-Ras GTPases. Does not interact with cAMP

Curated tissue note. Isoform 3 has highest expression levels in the brain, heart, liver, lung and placenta and is barely detectable in skeletal muscle, kidney and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TEU7 | HPA portal (live; not the pinned values)

RBL1 | Unsafe / Protein

ENSG00000080839

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / basal cells: Medium (Enhanced); bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / goblet cells: Medium (Enhanced); caudate / neuronal cells: Low (Enhanced); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 206/207 (99.5%)

Cancer IHC. melanoma: 12/12 (100.0%); urothelial cancer: 12/12 (100.0%)

Biology. Key regulator of entry into cell division (PubMed:17671431). Directly involved in heterochromatin formation by maintaining overall chromatin structure and, in particular, that of constitutive heterochromatin by stabilizing histone methylation (By similarity). Recruits and targets histone methyltransferases KMT5B and KMT5C, leading to epigenetic transcriptional repression (By similarity). Controls histone H4 'Lys-20' trimethylation (By ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P28749 | HPA portal (live; not the pinned values)

RBM12B | Unsafe / Protein

ENSG00000183808

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in molecular layer: Low (Uncertain); cerebellum / Purkinje cells: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. colorectal cancer: 8/12 (66.7%); liver cancer: 7/12 (58.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IXT5 | HPA portal (live; not the pinned values)

RBM14 | Unsafe / Protein

ENSG00000239306

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / glial cells: High (Supported); caudate / neuronal cells: Medium (Supported); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Isoform 1 may function as a nuclear receptor coactivator, enhancing transcription through other coactivators such as NCOA6 and CITED1. Isoform 2, functions as a transcriptional repressor, modulating transcriptional activities of coactivators including isoform 1, NCOA6 and CITED1 (PubMed:11443112). Regulates centriole biogenesis by suppressing the formation of aberrant centriolar protein complexes in the cytoplasm and thus preserving ... [full text in table]

Curated tissue note. Expressed in all tissues tested, including brain, heart, skeletal muscle, colon, thymus, spleen, kidney, liver, small intestine, placenta, lung and peripheral blood lymphocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96PK6 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

RBM34 | Unsafe / Protein

ENSG00000188739

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P42696 | HPA portal (live; not the pinned values)

RBM4 | Unsafe / Protein

ENSG00000173933

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); endometrial cancer: 10/10 (100.0%)

Biology. RNA-binding factor involved in multiple aspects of cellular processes like alternative splicing of pre-mRNA and translation regulation. Modulates alternative 5'-splice site and exon selection. Acts as a muscle cell differentiation-promoting factor. Activates exon skipping of the PTB pre-mRNA during muscle cell differentiation. Antagonizes the activity of the splicing factor PTBP1 to modulate muscle cell-specific exon selection of alpha ... [full text in table]

Curated tissue note. Expressed in the cerebellum. Expressed in neurons and glial cells, including layers II neurons in the frontal cortex and CA1 pyramidal neurons in the hippocampus. Expressed in heart, liver, pancreas, skeletal ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BWF3 | HPA portal (live; not the pinned values)

RBM48 | Unsafe / Protein

ENSG00000127993

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. lymphoma: 7/12 (58.3%); liver cancer: 4/12 (33.3%)

Biology. As a component of the minor spliceosome, involved in the splicing of U12-type introns in pre-mRNAs

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5RL73 | HPA portal (live; not the pinned values)

RBM7 | Unsafe / Protein

ENSG00000076053

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Supported); adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / basal cells: Low (Supported); bronchus / ciliated cells (cell body): Medium (Supported); bronchus / goblet cells: Medium (Supported); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. ovarian cancer: 8/12 (66.7%); stomach cancer: 8/12 (66.7%)

Biology. RNA-binding subunit of the trimeric nuclear exosome targeting (NEXT) complex, a complex that functions as an RNA exosome cofactor that directs a subset of non-coding short-lived RNAs for exosomal degradation (PubMed:25189701, PubMed:25525152, PubMed:25578728, PubMed:25852104, PubMed:27871484). NEXT is involved in surveillance and turnover of aberrant transcripts and non-coding RNAs (PubMed:25189701, PubMed:25852104, PubMed:27871484). ... [full text in table]

Curated tissue note. Ubiquitous

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y580 | HPA portal (live; not the pinned values)

RBPJL | Unsafe / Protein

ENSG00000124232

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. pancreas / pancreatic endocrine cells: Medium (Enhanced)

Cancer RNA. Pancreatic Adenocarcinoma (TCGA): 38/176 (21.6%); Pancreatic Adenocarcinoma (validation): 3/80 (3.8%); Ovary Serous Cystadenocarcinoma (TCGA): 3/349 (0.9%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Putative transcription factor, which cooperates with EBNA2 to activate transcription

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UBG7 | HPA portal (live; not the pinned values)

RDH10 | Unsafe / RNA

ENSG00000121039

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 41.8 nTPM (limit 10); adrenal gland: 6 nTPM (limit 2); amygdala: 5.4 nTPM (limit 2); basal ganglia: 5.3 nTPM (limit 2); bone marrow: 3.2 nTPM (limit 2); cerebral cortex: 6.8 nTPM (limit 2); choroid plexus: 15.5 nTPM (limit 2); colon: 16 nTPM (limit 6); duodenum: 31.5 nTPM (limit 2); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Retinol dehydrogenase with a clear preference for NADP. Converts all-trans-retinol to all-trans-retinal. Has no detectable activity towards 11-cis-retinol, 9-cis-retinol and 13-cis-retinol

Curated tissue note. Detected in retina, kidney, liver, small intestine, placenta, lung, heart and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZV5 | HPA portal (live; not the pinned values)

REC114 | Unsafe / RNA

ENSG00000183324

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. heart muscle: 4.5 nTPM (limit 2); heart muscle: 4.5 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 14/133 (10.5%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Liver Hepatocellular Carcinoma (TCGA): 2/362 (0.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Required for DNA double-strand breaks (DSBs) formation in unsynapsed regions during meiotic recombination (PubMed:38148155). Activates DSBs formation in unsynapsed regions, an essential step to ensure completion of synapsis (By similarity). Required for both spermatogenesis and oogenesis (PubMed:31704776, PubMed:38148155)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z4M0 | HPA portal (live; not the pinned values)

REM2 | Unsafe / RNA

ENSG00000139890

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 7.5 nTPM (limit 2); bone marrow: 9.7 nTPM (limit 2); cerebellum: 2.9 nTPM (limit 2); lung: 2.2 nTPM (limit 2); pituitary gland: 5.6 nTPM (limit 2); basal ganglia: 7.5 nTPM (baseline 2); bone marrow: 9.7 nTPM (baseline 2); cerebellum: 2.9 nTPM (baseline 2); lung: 2.2 nTPM (baseline ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 39/50 (78.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 29/176 (16.5%); Breast Invasive Carcinoma (TCGA): 51/1022 (5.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Binds GTP saturably and exhibits a low intrinsic rate of GTP hydrolysis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYK8 | HPA portal (live; not the pinned values)

REXO5 | Unsafe / Protein

ENSG00000005189

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / glandular cells: Low (Approved); kidney / cells in tubules: Low (Approved); rectum / glandular cells: Low (Approved); stomach 1 / glandular cells: Low (Approved); stomach 2 / glandular cells: Low (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 52/58 (89.7%); Prostate Adenocarcinoma (TCGA): 361/480 (75.2%); Colon Adenocarcinoma (validation): 230/486 (47.3%)

Cancer IHC. liver cancer: 6/12 (50.0%); pancreatic cancer: 2/11 (18.2%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96IC2 | HPA portal (live; not the pinned values)

RFESD | Unsafe / Protein

ENSG00000175449

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / cells in molecular layer: ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 29/50 (58.0%); Testicular Germ Cell Tumor (TCGA): 56/133 (42.1%); Glioblastoma Multiforme (TCGA): 51/141 (36.2%)

Cancer IHC. prostate cancer: 11/11 (100.0%); colorectal cancer: 11/12 (91.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TAC1 | HPA portal (live; not the pinned values)

RFX6 | Unsafe / Protein

ENSG00000185002

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / glandular cells: Medium (Approved); pancreas / pancreatic endocrine cells: High (Approved); rectum / glandular cells: Medium (Approved); small intestine / glandular cells: Low (Approved); stomach 1 / glandular cells: Medium (Approved); stomach 2 / glandular cells: Medium (Approved)

Cancer RNA. Pancreatic Adenocarcinoma (TCGA): 17/176 (9.7%); Breast Invasive Carcinoma (validation): 4/50 (8.0%); Prostate Adenocarcinoma (TCGA): 13/480 (2.7%)

Cancer IHC. endometrial cancer: 1/12 (8.3%); liver cancer: 1/12 (8.3%)

Biology. Transcription factor required to direct islet cell differentiation during endocrine pancreas development. Specifically required for the differentiation of 4 of the 5 islet cell types and for the production of insulin (PubMed:20148032, PubMed:25497100). Not required for pancreatic PP (polypeptide-producing) cells differentiation. Acts downstream of NEUROG3 and regulates the transcription factors involved in beta-cell maturation and ... [full text in table]

Curated tissue note. Expressed in pancreas (PubMed:25497100). Expressed in pancreatic beta-cells (insulin-positive cells) and alpha-cells (glucagon-positive cells) (at protein level). Specifically expressed in pancreas, small ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8HWS3 | HPA portal (live; not the pinned values)

RFX7 | Unsafe / Protein

ENSG00000181827

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / basal cells: Medium (Approved); bronchus / ciliated cells (cell body): Low (Approved); bronchus / goblet cells: Medium (Approved); caudate / glial cells: High (Approved); cerebellum / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. skin cancer: 3/12 (25.0%); liver cancer: 2/12 (16.7%)

Biology. Transcription factor (PubMed:29967452). Acts as a transcriptional activator by binding to promoter regions of target genes, such as PDCD4, PIK3IP1, MXD4, PNRC1, and RFX5 (PubMed:29967452, PubMed:34197623). Plays a role in natural killer (NK) cell maintenance and immunity (PubMed:29967452). May play a role in the process of ciliogenesis in the neural tube and neural tube closure (By similarity)

Curated tissue note. Widely expressed in many different tissue types including thymus and placenta, with high expression in brain (PubMed:18673564). Expressed in both inhibitory and excitatory neurons in cortex (PubMed:33658631)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q2KHR2 | HPA portal (live; not the pinned values)

RFX8 | Unsafe / RNA

ENSG00000196460

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 3.7 nTPM (limit 2); bone marrow: 3.7 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 24/50 (48.0%); Glioblastoma Multiforme (validation): 4/58 (6.9%); Pancreatic Adenocarcinoma (validation): 5/80 (6.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be a transcription factor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZV50 | HPA portal (live; not the pinned values)

RFXAP | Unsafe / RNA

ENSG00000133111

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.3 nTPM (limit 2); amygdala: 4.2 nTPM (limit 2); basal ganglia: 4.1 nTPM (limit 2); bone marrow: 5.9 nTPM (limit 2); cerebellum: 7 nTPM (limit 2); cerebral cortex: 6.5 nTPM (limit 2); duodenum: 3 nTPM (limit 2); heart muscle: 3.3 nTPM (limit 2); hippocampal formation: 4.1 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 44/50 (88.0%); Glioblastoma Multiforme (TCGA): 114/141 (80.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Part of the RFX complex that binds to the X-box of MHC II promoters

Curated tissue note. Ubiquitous

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00287 | HPA portal (live; not the pinned values)

RGR | Unsafe / RNA

ENSG00000148604

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 5.4 nTPM (limit 2); basal ganglia: 6.6 nTPM (limit 2); cerebral cortex: 10.4 nTPM (limit 2); choroid plexus: 33.4 nTPM (limit 2); hypothalamus: 2.1 nTPM (limit 2); midbrain: 3.2 nTPM (limit 2); retina: 134.8 nTPM (limit 2); amygdala: 5.4 nTPM (baseline 2); basal ganglia: 6.6 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 22/141 (15.6%); Glioblastoma Multiforme (validation): 8/58 (13.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 6/176 (3.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Non-visual opsin that functions as a light-dependent facilitator of retinoid isomerization and trafficking in retinal pigment epithelium (RPE) and Mueller glia (PubMed:18474598). Participates in a photic visual cycle by complementing the classical enzymatic RPE65/RDH pathway for chromophore regeneration (By similarity). Binds all-trans-retinal via a conserved Schiff-base lysine and, upon illumination, promotes photoisomerization of ... [full text in table]

Curated tissue note. Preferentially expressed at high levels in the retinal pigment epithelium (RPE) and Mueller cells of the neural retina

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P47804 | HPA portal (live; not the pinned values)

RGS22 | Unsafe / RNA

ENSG00000132554

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 6.4 nTPM (limit 2); choroid plexus: 6.4 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 39/58 (67.2%); Breast Invasive Carcinoma (validation): 23/50 (46.0%); Breast Invasive Carcinoma (TCGA): 124/1022 (12.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Inhibits signal transduction by increasing the GTPase activity of G protein alpha subunits thereby driving them into their inactive GDP-bound form

Curated tissue note. Testis-specific. Expressed in Leydig cells and spermatogenic cells from the spermatogonia to spermatid stages (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NE09 | HPA portal (live; not the pinned values)

RGS7 | Unsafe / Protein

ENSG00000182901

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Medium (Enhanced); cerebellum / GLUC cells - cytoplasm/membrane: High (Enhanced); cerebellum / molecular layer - neuropil: Medium (Enhanced); cerebellum / molecular layer cells - cytoplasm/membrane: Medium (Enhanced); cerebellum / Purkinje cells - cytoplasm/membrane: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 37/58 (63.8%); Glioblastoma Multiforme (TCGA): 29/141 (20.6%); Pancreatic Adenocarcinoma (validation): 9/80 (11.2%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. GTPase activator component of the RGS7-GNB5 complex that regulates G protein-coupled receptor signaling cascades (PubMed:10521509, PubMed:10862767, PubMed:31189666). The RGS7-GNB5 complex acts as an inhibitor signal transduction by promoting the GTPase activity of G protein alpha subunits, such as GNAO1, thereby driving them into their inactive GDP-bound form (PubMed:10521509, PubMed:10862767). May play a role in synaptic vesicle ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P49802 | HPA portal (live; not the pinned values)

RGS8 | Unsafe / Protein

ENSG00000135824

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: High (Enhanced); cerebellum / Purkinje cells - cytoplasm/membrane: High (Enhanced); cerebellum / Purkinje cells - dendrites: Medium (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 29/58 (50.0%); Testicular Germ Cell Tumor (TCGA): 17/133 (12.8%); Thyroid Carcinoma (TCGA): 37/495 (7.5%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Regulates G protein-coupled receptor signaling cascades, including signaling via muscarinic acetylcholine receptor CHRM2 and dopamine receptor DRD2 (By similarity). Inhibits signal transduction by increasing the GTPase activity of G protein alpha subunits, thereby driving them into their inactive GDP-bound form (PubMed:18434541). Modulates the activity of potassium channels that are activated in response to DRD2 and CHRM2 signaling (By ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P57771 | HPA portal (live; not the pinned values)

RHOXF1 | Unsafe / RNA

ENSG00000101883

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 12.6 nTPM (limit 4); choroid plexus: 3.8 nTPM (limit 2); hypothalamus: 3.7 nTPM (limit 2); retina: 2.5 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 16/133 (12.0%); Breast Invasive Carcinoma (validation): 5/50 (10.0%); Ovary Serous Cystadenocarcinoma (TCGA): 16/349 (4.6%)

Cancer IHC. testis cancer: 1/11 (9.1%); breast cancer: 0/12 (0.0%)

Biology. Transcription factor maybe involved in reproductive processes. Modulates expression of target genes encoding proteins involved in processes relevant to spermatogenesis

Curated tissue note. Ovary, testis and epididymis. Also detected in the prostate and the mammary gland. Expressed in many tumor cell lines derived from acute lymphocytic leukemia, prostate, endometrial adenocarcinoma, melanoma, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NHV9 | HPA portal (live; not the pinned values)

RIBC1 | Unsafe / RNA

ENSG00000158423

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.8 nTPM (limit 2); amygdala: 2.5 nTPM (limit 2); basal ganglia: 3.8 nTPM (limit 2); choroid plexus: 27.4 nTPM (limit 2); hippocampal formation: 3.2 nTPM (limit 2); hypothalamus: 4 nTPM (limit 2); kidney: 3.1 nTPM (limit 2); lung: 3.2 nTPM (limit 2); midbrain: 2.4 nTPM (limit 2); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 49/50 (98.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 162/282 (57.4%); Uterine Corpus Endometrial Carcinoma (TCGA): 81/176 (46.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N443 | HPA portal (live; not the pinned values)

RIBC2 | Unsafe / Protein

ENSG00000128408

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (tip of cilia): High (Approved); caudate / neuronal cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); hippocampus / neuronal cells: Medium (Approved); nasopharynx / ciliated cells (tip of cilia): High (Approved)

Cancer RNA. Breast Invasive Carcinoma (validation): 41/50 (82.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 187/283 (66.1%); Uterine Corpus Endometrial Carcinoma (TCGA): 50/176 (28.4%)

Cancer IHC. breast cancer: 4/12 (33.3%); endometrial cancer: 2/11 (18.2%)

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia axoneme, which is required for motile cilia beating

Curated tissue note. Expressed in airway epithelial cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H4K1 | HPA portal (live; not the pinned values)

RIC1 | Unsafe / Protein

ENSG00000107036

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: Medium (Supported); bronchus / respiratory epithelial cells: Medium (Supported); caudate / glial cells: Low (Supported); cerebellum / synaptic glomeruli - core: Medium (Supported); cerebellum / white matter ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. prostate cancer: 12/12 (100.0%); colorectal cancer: 11/11 (100.0%)

Biology. The RIC1-RGP1 complex acts as a guanine nucleotide exchange factor (GEF), which activates RAB6A by exchanging bound GDP for free GTP, and may thereby be required for efficient fusion of endosome-derived vesicles with the Golgi compartment (PubMed:23091056). The RIC1-RGP1 complex participates in the recycling of mannose-6-phosphate receptors (PubMed:23091056). Required for phosphorylation and localization of GJA1 (PubMed:16112082). Is a ... [full text in table]

Curated tissue note. Present in kidney and various cell lines (at protein level). Widely expressed at low level

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4ADV7 | HPA portal (live; not the pinned values)

RIIAD1 | Unsafe / Protein

ENSG00000178796

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); colon / goblet cells: Low (Enhanced); duodenum / enterocytes - Microvilli: Medium (Enhanced); nasopharynx / ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 19/176 (10.8%); Rectum Adenocarcinoma (TCGA): 7/88 (8.0%); Pancreatic Adenocarcinoma (TCGA): 11/176 (6.2%)

Cancer IHC. breast cancer: 2/11 (18.2%); liver cancer: 1/9 (11.1%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NNX1 | HPA portal (live; not the pinned values)

RIMS1 | Unsafe / Protein

ENSG00000079841

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: High (Enhanced); cerebral cortex / glial cells: Medium (Enhanced); cerebral cortex / neuronal cells: High (Enhanced); cerebral cortex / neuropil: High (Enhanced); hippocampus / glial cells: Low (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 38/58 (65.5%); Breast Invasive Carcinoma (validation): 10/50 (20.0%); Kidney Chromophobe (TCGA): 9/64 (14.1%)

Cancer IHC. colorectal cancer: 2/12 (16.7%); stomach cancer: 2/12 (16.7%)

Biology. Rab effector involved in exocytosis (By similarity). May act as scaffold protein that regulates neurotransmitter release at the active zone. Essential for maintaining normal probability of neurotransmitter release and for regulating release during short-term synaptic plasticity (By similarity). Plays a role in dendrite formation by melanocytes (PubMed:23999003)

Curated tissue note. Expressed in melanocytes (PubMed:23999003). Detected in brain and retina (PubMed:23999003)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86UR5 | HPA portal (live; not the pinned values)

RNASE8 | Unsafe / RNA

ENSG00000173431

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. salivary gland: 53.2 nTPM (limit 6); salivary gland: 53.2 nTPM (baseline 6)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Has a low ribonuclease activity

Curated tissue note. Expressed prominently in the placenta and is not detected in any other tissues examined

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TDE3 | HPA portal (live; not the pinned values)

RNF103-CHMP3 | Unsafe / RNA

ENSG00000249884

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 4.1 nTPM (limit 2); cerebral cortex: 2.4 nTPM (limit 2); heart muscle: 4 nTPM (limit 2); skeletal muscle: 2.1 nTPM (limit 2); bone marrow: 4.1 nTPM (baseline 2); cerebral cortex: 2.4 nTPM (baseline 2); heart muscle: 4 nTPM (baseline 2); skeletal muscle: 2.1 nTPM (baseline 2)

Cancer RNA. Colon Adenocarcinoma (validation): 100/486 (20.6%); Rectum Adenocarcinoma (validation): 37/207 (17.9%); Kidney Chromophobe (TCGA): 3/64 (4.7%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Probable core component of the endosomal sorting required for transport complex III (ESCRT-III) which is involved in multivesicular bodies (MVBs) formation and sorting of endosomal cargo proteins into MVBs. MVBs contain intraluminal vesicles (ILVs) that are generated by invagination and scission from the limiting membrane of the endosome and mostly are delivered to lysosomes enabling degradation of membrane proteins, such as stimulated ... [full text in table]

Curated tissue note. Widely expressed. Expressed in heart, brain, placenta, lung, liver, skeletal muscle, kidney and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y3E7 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

RNF138 | Unsafe / RNA

ENSG00000134758

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 13.2 nTPM (limit 10); adrenal gland: 7.1 nTPM (limit 2); amygdala: 6.9 nTPM (limit 2); basal ganglia: 6.2 nTPM (limit 2); bone marrow: 94.8 nTPM (limit 2); cerebellum: 8.4 nTPM (limit 2); cerebral cortex: 9.8 nTPM (limit 2); choroid plexus: 4.2 nTPM (limit 2); colon: 22.7 nTPM ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. E3 ubiquitin-protein ligase involved in DNA damage response by promoting DNA resection and homologous recombination (PubMed:26502055, PubMed:26502057). Recruited to sites of double-strand breaks following DNA damage and specifically promotes double-strand break repair via homologous recombination (PubMed:26502055, PubMed:26502057). Two different, non-exclusive, mechanisms have been proposed. According to a report, regulates the choice ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WVD3 | HPA portal (live; not the pinned values)

RNF157 | Unsafe / RNA

ENSG00000141576

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 13.1 nTPM (limit 10); adrenal gland: 3.2 nTPM (limit 2); amygdala: 46.2 nTPM (limit 2); basal ganglia: 41.9 nTPM (limit 2); bone marrow: 2.1 nTPM (limit 2); cerebellum: 23.8 nTPM (limit 2); cerebral cortex: 75 nTPM (limit 2); choroid plexus: 9.8 nTPM (limit 2); colon: 9.5 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 137/141 (97.2%); Colon Adenocarcinoma (validation): 429/486 (88.3%)

Cancer IHC. prostate cancer: 12/12 (100.0%); breast cancer: 11/11 (100.0%)

Biology. E3 ubiquitin ligase that ubiquitinates APBB1 for its degradation by the proteasome and thus prevents apoptosis and promotes survival of neurons (PubMed:25342469). Has a dual role in neurons as it is also required for dendrite growth and maintenance for which its ligase activity is not critical (PubMed:25342469). May act as a scaffold molecule to regulate this process (PubMed:25342469). Acts as a downstream effector of the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96PX1 | HPA portal (live; not the pinned values)

RNF169 | Unsafe / RNA

ENSG00000166439

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.8 nTPM (limit 2); bone marrow: 14.3 nTPM (limit 2); cerebellum: 2.6 nTPM (limit 2); cerebral cortex: 3.7 nTPM (limit 2); choroid plexus: 3.7 nTPM (limit 2); duodenum: 3.9 nTPM (limit 2); esophagus: 9.8 nTPM (limit 6); heart muscle: 3.1 nTPM (limit 2); kidney: 4.5 nTPM (limit 2); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. urothelial cancer: 1/10 (10.0%); breast cancer: 1/11 (9.1%)

Biology. Probable E3 ubiquitin-protein ligase that acts as a regulator of double-strand breaks (DSBs) repair following DNA damage. Functions in a non-canonical fashion to harness RNF168-mediated protein recruitment to DSB-containing chromatin, thereby contributing to regulation of DSB repair pathway utilization (PubMed:22492721, PubMed:30773093). Once recruited to DSB repair sites by recognizing and binding ubiquitin catalyzed by RNF168, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCN4 | HPA portal (live; not the pinned values)

RNF17 | Unsafe / Protein

ENSG00000132972

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 69/133 (51.9%); Liver Hepatocellular Carcinoma (validation): 7/231 (3.0%); Breast Invasive Carcinoma (validation): 1/50 (2.0%)

Cancer IHC. breast cancer: 11/12 (91.7%); testis cancer: 11/12 (91.7%)

Biology. Seems to be involved in regulation of transcriptional activity of MYC. In vitro, inhibits DNA-binding activity of Mad-MAX heterodimers. Can recruit Mad transcriptional repressors (MXD1, MXD3, MXD4 and MXI1) to the cytoplasm. May be involved in spermiogenesis (By similarity)

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BXT8 | HPA portal (live; not the pinned values)

RNF183 | Unsafe / RNA

ENSG00000165188

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 11.2 nTPM (limit 2); salivary gland: 6.6 nTPM (limit 6); kidney: 11.2 nTPM (baseline 2); salivary gland: 6.6 nTPM (baseline 6)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 121/176 (68.8%); Kidney Renal Clear Cell Carcinoma (validation): 52/100 (52.0%); Breast Invasive Carcinoma (validation): 26/50 (52.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Acts as an E3 ubiquitin ligase catalyzing the covalent attachment of ubiquitin moieties onto substrate proteins (PubMed:29507230). Triggers apoptosis in response to prolonged ER stress by mediating the polyubiquitination and subsequent proteasomal degradation of BCL2L1 (PubMed:29507230). May collaborate with FATE1 to restrain BIK protein levels thus regulating apoptotic signaling (PubMed:26567849). Mediates the ubiquitination and ... [full text in table]

Curated tissue note. Kidney and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96D59 | HPA portal (live; not the pinned values)

RNF212B | Unsafe / RNA

ENSG00000215277

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 25 nTPM (limit 2); kidney: 25 nTPM (baseline 2)

Cancer RNA. Kidney Chromophobe (TCGA): 5/64 (7.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 7/282 (2.5%); Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%)

Cancer IHC. pancreatic cancer: 1/10 (10.0%); cervical cancer: 1/11 (9.1%)

Biology. Ubiquitin E3 ligase that acts as a crucial factor for crossing-over (CO) formation during meiosis. Essential for normal prophase I progression and for ensuring appropriate CO designation in meiosis. Recruits key components of the cross-over machinery either directly ou indirectly, leading to the activation of the MutL-gamma complex. The function of RNF212B in CO designation is dependent on its catalytic activity

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A8MTL3 | HPA portal (live; not the pinned values)

RNF32 | Unsafe / RNA

ENSG00000105982

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.5 nTPM (limit 2); basal ganglia: 2.5 nTPM (limit 2); hippocampal formation: 2.3 nTPM (limit 2); hypothalamus: 2.3 nTPM (limit 2); midbrain: 2.2 nTPM (limit 2); retina: 2.7 nTPM (limit 2); spinal cord: 3.4 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 38/50 (76.0%); Rectum Adenocarcinoma (validation): 140/207 (67.6%); Colon Adenocarcinoma (validation): 284/486 (58.4%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. May play a role in sperm formation

Curated tissue note. Highly expressed in testis, less abundant in ovary

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0A6 | HPA portal (live; not the pinned values)

RNF6 | Unsafe / Protein

ENSG00000127870

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebral cortex / neuropil: High (Approved); colon / glandular cells: Medium (Approved); duodenum / glandular cells: Medium (Approved); ... [full text in table]

Cancer RNA. Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. skin cancer: 12/12 (100.0%); melanoma: 11/11 (100.0%)

Biology. E3 ubiquitin-protein ligase mediating 'Lys-48'-linked polyubiquitination of LIMK1 and its subsequent targeting to the proteasome for degradation (By similarity). Negatively regulates axonal outgrowth through regulation of the LIMK1 turnover (By similarity). Mediates 'Lys-6' and 'Lys-27'-linked polyubiquitination of AR/androgen receptor thereby modulating its transcriptional activity (PubMed:19345326). May also bind DNA and function as ... [full text in table]

Curated tissue note. Weakly expressed in peripheral blood, spleen, prostate, testis and ovary (PubMed:19345326). According to a report, it is preferentially expressed in testis and ovary and hardly detected in other tissues ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y252 | HPA portal (live; not the pinned values)

RNFT2 | Unsafe / Protein

ENSG00000135119

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / Purkinje cells: Medium (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); hippocampus / neuronal cells: Medium (Enhanced); lymph node / non-germinal center cells: Low (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Colon Adenocarcinoma (validation): 341/486 (70.2%); Breast Invasive Carcinoma (validation): 33/50 (66.0%)

Cancer IHC. pancreatic cancer: 1/8 (12.5%); breast cancer: 0/9 (0.0%)

Biology. E3 ubiquitin-protein ligase that negatively regulates IL3-dependent cellular responses through IL3RA ubiquitination and degradation by the proteasome, having an anti-inflammatory effect

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96EX2 | HPA portal (live; not the pinned values)

ROPN1 | Unsafe / High RNA

ENSG00000065371

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 32.9 nTPM; salivary gland/serous glandular cells; hpa_single_cell_tissue; protected; 0.5949 TPM; blood/TREG_NAIVE; dice_treg_naive; vital

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 81/99 (81.8%); Breast Invasive Carcinoma (validation): 15/50 (30.0%); Breast Invasive Carcinoma (TCGA): 160/1022 (15.7%)

Cancer IHC. colorectal cancer: 2/11 (18.2%); liver cancer: 1/9 (11.1%)

Biology. Important for male fertility. With ROPN1L, involved in fibrous sheath integrity and sperm motility, plays a role in PKA-dependent signaling processes required for spermatozoa capacitation

Curated tissue note. Testis specific in adult. Overexpressed in hematologic tumor cells

Reconsideration. Review the underlying cluster and gene-specific reads with reliable local protein evidence before reconsideration.

Therapeutic evidence. Clinical family label; locus unresolved.

UniProt Q9HAT0 | Trial/source record | HPA portal (live; not the pinned values)

ROPN1B | Unsafe / RNA

ENSG00000114547

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. 36.8 nTPM; heart muscle/cardiomyocytes; hpa_single_cell_tissue; vital; 1.624 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 95/99 (96.0%); Breast Invasive Carcinoma (validation): 19/50 (38.0%); Breast Invasive Carcinoma (TCGA): 181/1022 (17.7%)

Cancer IHC. colorectal cancer: 2/11 (18.2%); liver cancer: 1/9 (11.1%)

Biology. Important for male fertility. With ROPN1L, involved in fibrous sheath integrity and sperm motility, plays a role in PKA-dependent signaling processes required for spermatozoa capacitation

Therapeutic evidence. Clinical family label; locus unresolved.

UniProt Q9BZX4 | Trial/source record | HPA portal (live; not the pinned values)

ROPN1L | Unsafe / Protein

ENSG00000145491

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Low (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (tip of cilia): Medium (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 22/50 (44.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 45/176 (25.6%); Ovary Serous Cystadenocarcinoma (TCGA): 73/349 (20.9%)

Cancer IHC. ovarian cancer: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. Functions as part of axonemal radial spoke complexes that play an important part in the motility of sperm and cilia. Important for male fertility. With ROPN1, involved in fibrous sheath integrity and sperm motility, plays a role in PKA-dependent signaling processes required for spermatozoa capacitation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96C74 | HPA portal (live; not the pinned values)

RPA4 | Unsafe / Protein

ENSG00000204086

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 4/58 (6.9%); Pancreatic Adenocarcinoma (validation): 3/80 (3.8%); Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); lung cancer: 12/12 (100.0%)

Biology. As part of the alternative replication protein A complex, aRPA, binds single-stranded DNA and probably plays a role in DNA repair. Compared to the RPA2-containing, canonical RPA complex, may not support chromosomal DNA replication and cell cycle progression through S-phase. The aRPA may not promote efficient priming by DNA polymerase alpha but could support DNA polymerase delta synthesis in the presence of PCNA and replication factor C ... [full text in table]

Curated tissue note. Preferentially expressed in placental and colon mucosa. Widely expressed at intermediate or lower levels

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13156 | HPA portal (live; not the pinned values)

RPAP3 | Unsafe / Protein

ENSG00000005175

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Enhanced); adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: High (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. lymphoma: 12/12 (100.0%); melanoma: 12/12 (100.0%)

Biology. Forms an interface between the RNA polymerase II enzyme and chaperone/scaffolding protein, suggesting that it is required to connect RNA polymerase II to regulators of protein complex formation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H6T3 | HPA portal (live; not the pinned values)

RPGR | Unsafe / Protein

ENSG00000156313

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / ciliated cells (cilia axoneme): Medium (Approved); bronchus / ciliated cells (ciliary rootlets): High (Approved); bronchus / ciliated cells (tip of cilia): Medium (Approved); caudate ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 48/50 (96.0%); Pancreatic Adenocarcinoma (validation): 71/80 (88.8%)

Cancer IHC. liver cancer: 11/11 (100.0%); skin cancer: 11/11 (100.0%)

Biology. Acts as a guanine-nucleotide releasing factor (GEF) for RAB8A and RAB37 by promoting the conversion of inactive RAB-GDP to the active form RAB-GTP (PubMed:20631154). GEF activity towards RAB8A may facilitate ciliary trafficking by modulating ciliary intracellular localization of RAB8A (PubMed:20631154). GEF activity towards RAB37 maintains autophagic homeostasis and retinal function (By similarity). Involved in photoreceptor integrity ... [full text in table]

Curated tissue note. Heart, brain, placenta, lung, liver, muscle, kidney, retina, pancreas and fetal retinal pigment epithelium. Isoform 3 is found only in the retina. Colocalizes with RPGRIP1 in the outer segment of rod ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92834 | HPA portal (live; not the pinned values)

RPGRIP1 | Unsafe / Protein

ENSG00000092200

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: High (Supported)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 26/133 (19.5%); Breast Invasive Carcinoma (validation): 8/50 (16.0%); Lung Adenocarcinoma (validation): 2/105 (1.9%)

Cancer IHC. melanoma: 1/12 (8.3%); breast cancer: 0/9 (0.0%)

Biology. May function as scaffolding protein. Required for normal location of RPGR at the connecting cilium of photoreceptor cells. Required for normal disk morphogenesis and disk organization in the outer segment of photoreceptor cells and for survival of photoreceptor cells

Curated tissue note. Strong expression in retina, with weaker expression in testis. Expressed in other neurons such as amacrine cells. Colocalizes with RGPR in the outer segment of rod photoreceptors and cone outer segments

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96KN7 | HPA portal (live; not the pinned values)

RPL10L | Unsafe / High RNA

ENSG00000165496

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 41.3 nTPM; bronchus/smooth muscle cells; hpa_single_cell_tissue; vital; 1.857 TPM; blood/M2; dice_m2; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 75/133 (56.4%); Liver Hepatocellular Carcinoma (validation): 13/231 (5.6%); Liver Hepatocellular Carcinoma (TCGA): 12/362 (3.3%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Testis-specific component of the ribosome, which is required for the transition from prophase to metaphase in male meiosis I (By similarity). Compensates for the inactivated X-linked RPL10 paralog during spermatogenesis (PubMed:12490704). The ribosome is a large ribonucleoprotein complex responsible for the synthesis of proteins in the cell (PubMed:23636399, PubMed:25901680, PubMed:32669547). The male germ cell-specific ribosome ... [full text in table]

Curated tissue note. Almost testis-specific (PubMed:12490704, PubMed:19333399, PubMed:28502657). Also expressed in pre- and postmenopausal ovary (PubMed:19333399)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96L21 | HPA portal (live; not the pinned values)

RPL39L | Unsafe / Protein

ENSG00000163923

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / glandular cells: Low (Approved); duodenum / glandular cells: Low (Approved); heart muscle / cardiomyocytes: Low (Approved); kidney / cells in tubules: Low (Approved); liver / hepatocytes: Low (Approved); rectum / glandular cells: Low (Approved); salivary gland / glandular cells: Low ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. liver cancer: 3/12 (25.0%); breast cancer: 1/12 (8.3%)

Biology. Male germ cell-specific component of the ribosome, which is required for the formation of sperm and male fertility. Replaces the RPL39 paralog in the ribosome of male germ cells. The ribosome is a large ribonucleoprotein complex responsible for the synthesis of proteins in the cell. The male germ cell-specific ribosome displays a ribosomal polypeptide exit tunnel of distinct size and charge states compared with the classical ribosome. ... [full text in table]

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96EH5 | HPA portal (live; not the pinned values)

RPP14 | Unsafe / RNA

ENSG00000163684

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 5.5 nTPM (limit 2); amygdala: 4 nTPM (limit 2); basal ganglia: 5 nTPM (limit 2); bone marrow: 4.7 nTPM (limit 2); cerebellum: 6.4 nTPM (limit 2); cerebral cortex: 5.3 nTPM (limit 2); choroid plexus: 3.5 nTPM (limit 2); colon: 6.6 nTPM (limit 6); duodenum: 6.6 nTPM (limit 2); heart ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Liver Hepatocellular Carcinoma (validation): 228/231 (98.7%); Colon Adenocarcinoma (validation): 478/486 (98.4%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); liver cancer: 9/11 (81.8%)

Biology. Component of ribonuclease P, a ribonucleoprotein complex that generates mature tRNA molecules by cleaving their 5'-ends

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95059 | HPA portal (live; not the pinned values)

RPP38 | Unsafe / Protein

ENSG00000152464

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / glial cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); colon / glandular cells: Low (Approved); duodenum / ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%)

Cancer IHC. stomach cancer: 9/12 (75.0%); lung cancer: 8/12 (66.7%)

Biology. Component of ribonuclease P, a ribonucleoprotein complex that generates mature tRNA molecules by cleaving their 5'-ends (PubMed:10444065, PubMed:30454648, PubMed:9037013, PubMed:9630247). Also a component of the MRP ribonuclease complex, which cleaves pre-rRNA sequences (PubMed:28115465)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P78345 | HPA portal (live; not the pinned values)

RPRD1A | Unsafe / Protein

ENSG00000141425

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: Low (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / glial cells: Medium (Supported); caudate / neuronal cells: High (Supported); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); cervical cancer: 11/12 (91.7%)

Biology. Interacts with phosphorylated C-terminal heptapeptide repeat domain (CTD) of the largest RNA polymerase II subunit POLR2A, and participates in dephosphorylation of the CTD by RPAP2. May act as a negative regulator of cyclin-D1 (CCND1) and cyclin-E (CCNE1) in the cell cycle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96P16 | HPA portal (live; not the pinned values)

RSPH10B | Unsafe / Protein

ENSG00000155026

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Supported); bronchus / ciliated cells (tip of cilia): High (Supported)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 100/176 (56.8%); Prostate Adenocarcinoma (TCGA): 115/480 (24.0%); Testicular Germ Cell Tumor (TCGA): 31/133 (23.3%)

Cancer IHC. liver cancer: 2/11 (18.2%); breast cancer: 1/12 (8.3%)

Biology. May function as part of the axonemal radial spoke complex 3 (RS3). Radial spoke complexes are important for ciliary motility

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt B2RC85 | UniProt P0C881 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

RSPH10B2 | Unsafe / Protein

ENSG00000169402

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Supported); bronchus / ciliated cells (tip of cilia): High (Supported)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 12/133 (9.0%); Pancreatic Adenocarcinoma (validation): 6/80 (7.5%); Glioblastoma Multiforme (validation): 1/58 (1.7%)

Cancer IHC. liver cancer: 2/11 (18.2%); breast cancer: 1/12 (8.3%)

Biology. May function as part of the axonemal radial spoke complex 3 (RS3). Radial spoke complexes are important for ciliary motility

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt B2RC85 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

RTEL1 | Unsafe / RNA

ENSG00000258366

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 6.8 nTPM (limit 2); basal ganglia: 3.1 nTPM (limit 2); bone marrow: 5.2 nTPM (limit 2); cerebellum: 14.5 nTPM (limit 2); cerebral cortex: 5.9 nTPM (limit 2); choroid plexus: 2.4 nTPM (limit 2); colon: 8.3 nTPM (limit 6); duodenum: 3.2 nTPM (limit 2); heart muscle: 3.8 nTPM (limit 2); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%); Breast Invasive Carcinoma (validation): 47/50 (94.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. A probable ATP-dependent DNA helicase implicated in telomere-length regulation, DNA repair and the maintenance of genomic stability. Acts as an anti-recombinase to counteract toxic recombination and limit crossover during meiosis. Regulates meiotic recombination and crossover homeostasis by physically dissociating strand invasion events and thereby promotes noncrossover repair by meiotic synthesis dependent strand annealing (SDSA) as ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NZ71 | HPA portal (live; not the pinned values)

S100A7A | Unsafe / Protein

ENSG00000184330

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. esophagus / squamous epithelial cells: Medium (Supported); oral mucosa / squamous epithelial cells: Low (Supported); skin 1 / keratinocytes: High (Supported); skin 1 / Langerhans: Low (Supported); skin 2 / epidermal cells: High (Supported)

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 292/492 (59.3%); Skin Cuteneous Melanoma (TCGA): 32/99 (32.3%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 75/283 (26.5%)

Cancer IHC. skin cancer: 5/10 (50.0%); cervical cancer: 5/12 (41.7%)

Biology. May be involved in epidermal differentiation and inflammation and might therefore be important for the pathogenesis of psoriasis and other diseases

Curated tissue note. Overexpressed in psoriasis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86SG5 | HPA portal (live; not the pinned values)

SAC3D1 | Unsafe / Protein

ENSG00000168061

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); ... [full text in table]

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 348/349 (99.7%); Glioblastoma Multiforme (TCGA): 140/141 (99.3%); Ovary Serous Cystadenocarcinoma (validation): 79/81 (97.5%)

Cancer IHC. melanoma: 10/11 (90.9%); ovarian cancer: 10/11 (90.9%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

HPA portal (live; not the pinned values)

SAG | Unsafe / Protein

ENSG00000130561

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / photoreceptor cells: High (Enhanced)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 6/80 (7.5%); Breast Invasive Carcinoma (validation): 1/50 (2.0%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. skin cancer: 1/11 (9.1%); breast cancer: 0/12 (0.0%)

Biology. Involved in the regulation of phototransduction, playing a key role in signal termination after rhodopsin (RHO) has been activated by light in rod photoreceptors. Binds to photoactivated and phosphorylated RHO, preventing its interaction with transducin (GNAT1) and terminating transducin-mediated opsin signaling (By similarity). May play a role in preventing light-dependent degeneration of retinal photoreceptor cells (PubMed:9565049)

Curated tissue note. Detected in retina, in the proximal portion of the outer segment of rod photoreceptor cells (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P10523 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

SAMD15 | Unsafe / Protein

ENSG00000100583

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Enhanced); adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: High (Enhanced); bronchus / respiratory epithelial cells: High (Enhanced); caudate / glial cells: High (Enhanced); caudate / neuronal cells: Medium (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 53/58 (91.4%); Uterine Corpus Endometrial Carcinoma (TCGA): 45/176 (25.6%); Breast Invasive Carcinoma (TCGA): 170/1022 (16.6%)

Cancer IHC. cervical cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P1V8 | HPA portal (live; not the pinned values)

SAMD3 | Unsafe / RNA

ENSG00000164483

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.2 nTPM (limit 2); bone marrow: 4.2 nTPM (limit 2); hippocampal formation: 2.4 nTPM (limit 2); lung: 3.2 nTPM (limit 2); lymph node: 10.1 nTPM (limit 6); pituitary gland: 4.7 nTPM (limit 2); adrenal gland: 2.2 nTPM (baseline 2); bone marrow: 4.2 nTPM (baseline 2); hippocampal ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 33/50 (66.0%); Kidney Renal Clear Cell Carcinoma (validation): 29/100 (29.0%); Lung Adenocarcinoma (validation): 19/105 (18.1%)

Cancer IHC. urothelial cancer: 9/12 (75.0%); liver cancer: 8/11 (72.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N6K7 | HPA portal (live; not the pinned values)

SAMD7 | Unsafe / RNA

ENSG00000187033

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 130 nTPM (limit 2); retina: 130 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 2/176 (1.1%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Ovary Serous Cystadenocarcinoma (TCGA): 1/349 (0.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of a Polycomb group (PcG) multiprotein PRC1-like complex, essential for establishing rod photoreceptor cell identity and function by silencing nonrod gene expression in developing rod photoreceptor cells. Via its association with the PRC1-like complex, promotes epigenetic repressive marks H3K27me3 and H2AK119ub marks in nonrod genes, silencing their transcription. Represses Crx-controlled photoreceptor-specific gene expression

Curated tissue note. Expressed in the retina (at protein level) (PubMed:26887858). Expressed in the retinal inner and outer nuclear layers (PubMed:38272031)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z3H4 | HPA portal (live; not the pinned values)

SARS2 | Unsafe / Protein

ENSG00000104835

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / cells in molecular layer: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Breast Invasive Carcinoma (validation): 50/50 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 486/489 (99.4%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); ovarian cancer: 11/12 (91.7%)

Biology. Catalyzes the attachment of serine to tRNA(Ser). Is also probably able to aminoacylate tRNA(Sec) with serine, to form the misacylated tRNA L-seryl-tRNA(Sec), which will be further converted into selenocysteinyl-tRNA(Sec)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NP81 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

SASS6 | Unsafe / Protein

ENSG00000156876

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); caudate / glial cells: Medium (Uncertain); cerebral cortex / endothelial cells: Medium (Uncertain); cerebral cortex / glial cells: Medium (Uncertain); cerebral cortex / neuropil: Low (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 47/50 (94.0%); Colon Adenocarcinoma (validation): 343/486 (70.6%)

Cancer IHC. lung cancer: 5/11 (45.5%); liver cancer: 5/12 (41.7%)

Biology. Central scaffolding component of the centrioles ensuring their 9-fold symmetry (By similarity). Required for centrosome biogenesis and duplication: required both for mother-centriole-dependent centriole duplication and deuterosome-dependent centriole amplification in multiciliated cells (PubMed:15665853, PubMed:16244668, PubMed:17681131). Not required for centriole formation in embryonic stem cells but necessary to maintain centriole ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6UVJ0 | HPA portal (live; not the pinned values)

SAXO1 | Unsafe / Protein

ENSG00000155875

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced); nasopharynx / respiratory epithelial cells: Low (Enhanced)

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Breast Invasive Carcinoma (TCGA): 0/1022 (0.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May play a role in the regulation of cilium length. Stabilizes microtubules at low temperature

Curated tissue note. Widely expressed, with highest levels in testis. Expressed in mature spermatozoa (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYX7 | HPA portal (live; not the pinned values)

SAXO4 | Unsafe / Protein

ENSG00000162148

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); colon / enterocytes - Microvilli: Medium (Enhanced); colon / mucosal lymphoid cells: Low (Enhanced); duodenum / enterocytes - Microvilli: Low (Enhanced); nasopharynx / ciliated ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 107/141 (75.9%); Rectum Adenocarcinoma (validation): 130/207 (62.8%); Colon Adenocarcinoma (validation): 249/486 (51.2%)

Cancer IHC. renal cancer: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z5V6 | HPA portal (live; not the pinned values)

SCAI | Unsafe / Protein

ENSG00000173611

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: High (Approved); cerebellum / cells in molecular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Colon Adenocarcinoma (validation): 364/486 (74.9%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); prostate cancer: 10/11 (90.9%)

Biology. Tumor suppressor which functions to suppress MRTFA-induced SRF transcriptional activity. May function in the RHOA-DIAPH1 signal transduction pathway and regulate cell migration through transcriptional regulation of ITGB1

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N9R8 | HPA portal (live; not the pinned values)

SCML2 | Unsafe / Protein

ENSG00000102098

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Low (Enhanced); colon / glandular cells: Medium (Enhanced); esophagus / squamous epithelial cells: Medium (Enhanced); kidney / cells in tubules: Low (Enhanced); lung / alveolar cells: Low (Enhanced); oral mucosa / squamous epithelial cells: Medium (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 45/58 (77.6%); Testicular Germ Cell Tumor (TCGA): 83/133 (62.4%); Breast Invasive Carcinoma (validation): 23/50 (46.0%)

Cancer IHC. testis cancer: 1/11 (9.1%); colorectal cancer: 1/12 (8.3%)

Biology. Putative Polycomb group (PcG) protein. PcG proteins act by forming multiprotein complexes, which are required to maintain the transcriptionally repressive state of homeotic genes throughout development (By similarity)

Curated tissue note. Highly expressed in placenta, thymus and testis. Detected at lower levels in brain, liver, skeletal muscle, pancreas and ovary

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UQR0 | HPA portal (live; not the pinned values)

SDE2 | Unsafe / Protein

ENSG00000143751

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. pancreatic cancer: 8/11 (72.7%); breast cancer: 8/12 (66.7%)

Biology. Inhibits translesion DNA synthesis by preventing monoubiquitination of PCNA, this is necessary to counteract damage due to ultraviolet light-induced replication stress (PubMed:27906959). SDE2 is cleaved following PCNA binding, and its complete degradation is necessary to allow S-phase progression following DNA damage (PubMed:27906959) Plays a role in pre-mRNA splicing by facilitating excision of relatively short introns featuring weak ... [full text in table]

Curated tissue note. Ubiquitously expressed; enriched in brain, lung and liver

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6IQ49 | HPA portal (live; not the pinned values)

SDHAF2 | Unsafe / Protein

ENSG00000167985

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / glial cells: Low (Supported); caudate / neuronal cells: High (Supported); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Plays an essential role in the assembly of succinate dehydrogenase (SDH), an enzyme complex (also referred to as respiratory complex II) that is a component of both the tricarboxylic acid (TCA) cycle and the mitochondrial electron transport chain, and which couples the oxidation of succinate to fumarate with the reduction of ubiquinone (coenzyme Q) to ubiquinol. Required for flavinylation (covalent attachment of FAD) of the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NX18 | HPA portal (live; not the pinned values)

SDK2 | Unsafe / RNA

ENSG00000069188

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. spinal cord: 2.2 nTPM (limit 2); spinal cord: 2.2 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 20/50 (40.0%); Lung Squamous Cell Carcinoma (TCGA): 171/489 (35.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 167/492 (33.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Adhesion molecule that promotes lamina-specific synaptic connections in the retina and is specifically required for the formation of neuronal circuits that detect motion. Acts by promoting formation of synapses between two specific retinal cell types: the retinal ganglion cells W3B-RGCs and the excitatory amacrine cells VG3-ACs. Formation of synapses between these two cells plays a key role in detection of motion. Promotes synaptic ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q58EX2 | HPA portal (live; not the pinned values)

SEC16B | Unsafe / Protein

ENSG00000120341

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); duodenum / endocrine cells: Medium (Enhanced); duodenum / enterocytes: Medium (Enhanced); pancreas / exocrine glandular cells: Low (Enhanced); small intestine / endocrine cells: Medium (Enhanced); small intestine / enterocytes: Medium (Enhanced); ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 228/231 (98.7%); Kidney Renal Clear Cell Carcinoma (validation): 93/100 (93.0%); Rectum Adenocarcinoma (validation): 189/207 (91.3%)

Cancer IHC. breast cancer: 0/9 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Plays a role in the organization of the endoplasmic reticulum exit sites (ERES), also known as transitional endoplasmic reticulum (tER). Required for secretory cargo traffic from the endoplasmic reticulum to the Golgi apparatus (PubMed:17192411, PubMed:21768384, PubMed:22355596). Involved in peroxisome biogenesis. Regulates the transport of peroxisomal biogenesis factors PEX3 and PEX16 from the ER to peroxisomes (PubMed:21768384)

Curated tissue note. Ubiquitous

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96JE7 | HPA portal (live; not the pinned values)

SENP5 | Unsafe / RNA

ENSG00000119231

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 6.8 nTPM (limit 2); amygdala: 9.7 nTPM (limit 2); basal ganglia: 9.3 nTPM (limit 2); bone marrow: 15.6 nTPM (limit 2); cerebellum: 15.5 nTPM (limit 2); cerebral cortex: 11.5 nTPM (limit 2); choroid plexus: 6.7 nTPM (limit 2); colon: 9.9 nTPM (limit 6); duodenum: 5.7 nTPM (limit 2); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Protease that catalyzes two essential functions in the SUMO pathway: processing of full-length SUMO3 to its mature form and deconjugation of SUMO2 and SUMO3 from targeted proteins. Has weak proteolytic activity against full-length SUMO1 or SUMO1 conjugates. Required for cell division

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96HI0 | HPA portal (live; not the pinned values)

SENP8 | Unsafe / Protein

ENSG00000166192

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 459/486 (94.4%); Breast Invasive Carcinoma (validation): 45/50 (90.0%)

Cancer IHC. ovarian cancer: 12/12 (100.0%); breast cancer: 11/11 (100.0%)

Biology. Protease that catalyzes two essential functions in the NEDD8 pathway: processing of full-length NEDD8 to its mature form and deconjugation of NEDD8 from targeted proteins such as cullins or p53

Curated tissue note. Broadly expressed, with highest levels in kidney and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96LD8 | HPA portal (live; not the pinned values)

SEPSECS | Unsafe / RNA

ENSG00000109618

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.4 nTPM (limit 2); amygdala: 2.2 nTPM (limit 2); basal ganglia: 2.1 nTPM (limit 2); cerebellum: 3.4 nTPM (limit 2); duodenum: 4.7 nTPM (limit 2); heart muscle: 2.7 nTPM (limit 2); hippocampal formation: 2.5 nTPM (limit 2); hypothalamus: 2.2 nTPM (limit 2); kidney: 7.9 nTPM (limit ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 49/50 (98.0%); Pancreatic Adenocarcinoma (validation): 77/80 (96.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Converts O-phosphoseryl-tRNA(Sec) to selenocysteinyl-tRNA(Sec) required for selenoprotein biosynthesis

Curated tissue note. Primarily expressed in liver, pancreas, kidney and lung. Overexpressed in PHA-stimulated T-cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HD40 | HPA portal (live; not the pinned values)

SEPTIN12 | Unsafe / RNA

ENSG00000140623

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 3.1 nTPM (limit 2); cerebellum: 3.1 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 1/282 (0.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Filament-forming cytoskeletal GTPase (By similarity). Involved in the morphogenesis of sperm heads and the elongation of sperm tails probably implicating the association with alpha- and beta-tubulins (PubMed:24213608). Facilitates the formation of Septin filament structures with SEPTIN7, SEPTIN6, SEPTIN2 and SEPTIN4 at the sperm annulus (PubMed:25588830, PubMed:28346465). Correct formation of the annulus is required for development of ... [full text in table]

Curated tissue note. Widely expressed. Expressed in lymph node

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYM1 | HPA portal (live; not the pinned values)

SERINC4 | Unsafe / RNA

ENSG00000184716

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3.5 nTPM (limit 2); retina: 14.9 nTPM (limit 2); choroid plexus: 3.5 nTPM (baseline 2); retina: 14.9 nTPM (baseline 2)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 116/231 (50.2%); Breast Invasive Carcinoma (validation): 20/50 (40.0%); Prostate Adenocarcinoma (TCGA): 44/480 (9.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Incorporates a polar amino acid serine into membranes and facilitates the synthesis of two serine-derived lipids, phosphatidylserine and sphingolipids

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NH21 | HPA portal (live; not the pinned values)

SESN3 | Unsafe / RNA

ENSG00000149212

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 16.9 nTPM (limit 2); amygdala: 18.2 nTPM (limit 2); basal ganglia: 30.7 nTPM (limit 2); bone marrow: 17.7 nTPM (limit 2); cerebellum: 7.1 nTPM (limit 2); cerebral cortex: 20 nTPM (limit 2); choroid plexus: 9.6 nTPM (limit 2); duodenum: 6.3 nTPM (limit 2); esophagus: 8.6 nTPM (limit ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 140/141 (99.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May function as an intracellular leucine sensor that negatively regulates the TORC1 signaling pathway (PubMed:25263562). May also regulate the insulin-receptor signaling pathway through activation of TORC2 (By similarity). This metabolic regulator may also play a role in protection against oxidative and genotoxic stresses (By similarity). May prevent the accumulation of reactive oxygen species (ROS) through the alkylhydroperoxide ... [full text in table]

Curated tissue note. Widely expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P58005 | HPA portal (live; not the pinned values)

SFR1 | Unsafe / RNA

ENSG00000156384

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.3 nTPM (limit 2); amygdala: 5 nTPM (limit 2); basal ganglia: 5.2 nTPM (limit 2); bone marrow: 9.8 nTPM (limit 2); cerebellum: 3.4 nTPM (limit 2); cerebral cortex: 6.6 nTPM (limit 2); choroid plexus: 7.8 nTPM (limit 2); duodenum: 4.5 nTPM (limit 2); heart muscle: 7.3 nTPM (limit 2); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. melanoma: 4/11 (36.4%); liver cancer: 2/11 (18.2%)

Biology. Component of the SWI5-SFR1 complex, a complex required for double-strand break repair via homologous recombination (PubMed:21252223). Acts as a transcriptional modulator for ESR1 (PubMed:23874500)

Curated tissue note. Widely expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86XK3 | HPA portal (live; not the pinned values)

SGMS2 | Unsafe / Protein

ENSG00000164023

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: Medium (Supported); bronchus / basal cells: Medium (Supported); bronchus / ciliated cells (cell body): High (Supported); bronchus / ciliated cells (cilia axoneme): Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); stomach cancer: 11/11 (100.0%)

Biology. Sphingomyelin synthase that primarily contributes to sphingomyelin synthesis and homeostasis at the plasma membrane. Catalyzes the reversible transfer of phosphocholine moiety in sphingomyelin biosynthesis: in the forward reaction transfers phosphocholine head group of phosphatidylcholine (PC) on to ceramide (CER) to form ceramide phosphocholine (sphingomyelin, SM) and diacylglycerol (DAG) as by-product, and in the reverse reaction ... [full text in table]

Curated tissue note. Brain, heart, kidney, liver, muscle and stomach. Also expressed in a number of cell lines such as carcinoma HeLa cells, hepatoma Hep-G2 cells, and colon carcinoma Caco-2 cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NHU3 | HPA portal (live; not the pinned values)

SGO1 | Unsafe / Protein

ENSG00000129810

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / enterocytes: Medium (Enhanced); colon / goblet cells: Medium (Enhanced); duodenum / enterocytes: Medium (Enhanced); duodenum / goblet cells: Medium (Enhanced); esophagus / squamous epithelial cells: Low (Enhanced); lymph node / non-germinal center cells: Low (Enhanced); oral mucosa / ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 476/486 (97.9%); Rectum Adenocarcinoma (validation): 196/207 (94.7%); Testicular Germ Cell Tumor (TCGA): 109/133 (82.0%)

Cancer IHC. melanoma: 5/10 (50.0%); cervical cancer: 5/12 (41.7%)

Biology. Plays a central role in chromosome cohesion during mitosis by preventing premature dissociation of cohesin complex from centromeres after prophase, when most of cohesin complex dissociates from chromosomes arms. May act by preventing phosphorylation of the STAG2 subunit of cohesin complex at the centromere, ensuring cohesin persistence at centromere until cohesin cleavage by ESPL1/separase at anaphase. Essential for proper chromosome ... [full text in table]

Curated tissue note. Widely expressed. Highly expressed in testis. Expressed in lung, small intestine, breast, liver and placenta. Strongly overexpressed in 90% of breast cancers tested

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5FBB7 | HPA portal (live; not the pinned values)

SGO2 | Unsafe / Protein

ENSG00000163535

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / basal cells: Low (Enhanced); colon / enterocytes: Low (Enhanced); colon / goblet cells: Low (Enhanced); duodenum / endocrine cells: Medium (Enhanced); duodenum / goblet cells: Low (Enhanced); esophagus / squamous epithelial cells: Low (Enhanced); lymph node / non-germinal center cells: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 460/486 (94.7%); Rectum Adenocarcinoma (validation): 183/207 (88.4%)

Cancer IHC. cervical cancer: 2/12 (16.7%); colorectal cancer: 1/11 (9.1%)

Biology. Cooperates with PPP2CA to protect centromeric cohesin from separase-mediated cleavage in oocytes specifically during meiosis I. Has a crucial role in protecting REC8 at centromeres from cleavage by separase. During meiosis, protects centromeric cohesion complexes until metaphase II/anaphase II transition, preventing premature release of meiosis-specific REC8 cohesin complexes from anaphase I centromeres. Is thus essential for an ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q562F6 | HPA portal (live; not the pinned values)

SH2D7 | Unsafe / Protein

ENSG00000183476

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / endocrine cells: High (Enhanced); duodenum / endocrine cells: High (Enhanced); duodenum / enterocytes - Microvilli: Low (Enhanced); rectum / endocrine cells: High (Enhanced); small intestine / endocrine cells: High (Enhanced); small intestine / enterocytes - Microvilli: Low (Enhanced); ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 18/176 (10.2%); Pancreatic Adenocarcinoma (validation): 4/80 (5.0%); Pancreatic Adenocarcinoma (TCGA): 3/176 (1.7%)

Cancer IHC. pancreatic cancer: 7/11 (63.6%); skin cancer: 2/12 (16.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NKC9 | HPA portal (live; not the pinned values)

SHC4 | Unsafe / RNA

ENSG00000185634

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 8.5 nTPM (limit 2); basal ganglia: 10.2 nTPM (limit 2); cerebral cortex: 7.5 nTPM (limit 2); heart muscle: 3.2 nTPM (limit 2); hippocampal formation: 10.8 nTPM (limit 2); hypothalamus: 6.1 nTPM (limit 2); midbrain: 12.5 nTPM (limit 2); spinal cord: 15.4 nTPM (limit 2); amygdala: 8.5 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Skin Cuteneous Melanoma (TCGA): 72/99 (72.7%); Glioblastoma Multiforme (TCGA): 76/141 (53.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Activates both Ras-dependent and Ras-independent migratory pathways in melanomas. Contributes to the early phases of agrin-induced tyrosine phosphorylation of CHRNB1

Curated tissue note. Only expressed in melanomas. Weakly expressed in normal melanocytes and benign nevi. Highly expressed at the transition from radial growth phase to vertical growth phase and metastatic melanomas, when tumor ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6S5L8 | HPA portal (live; not the pinned values)

SHCBP1L | Unsafe / Protein

ENSG00000157060

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / distal tubules: Medium (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 3/283 (1.1%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/9 (0.0%)

Biology. Testis-specific spindle-associated factor that plays a role in spermatogenesis. In association with HSPA2, participates in the maintenance of spindle integrity during meiosis in male germ cells

Curated tissue note. Expressed in spermatocytes and elongating spermatids inside the seminiferous tubules (at protein level) (PubMed:24557841). Testis-specific (PubMed:11318611, PubMed:24557841)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BZQ2 | HPA portal (live; not the pinned values)

SHOC1 | Unsafe / RNA

ENSG00000165181

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.7 nTPM (limit 2); kidney: 2.2 nTPM (limit 2); adrenal gland: 3.7 nTPM (baseline 2); kidney: 2.2 nTPM (baseline 2)

Cancer RNA. Kidney Chromophobe (TCGA): 34/64 (53.1%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 20/283 (7.1%); Lung Adenocarcinoma (validation): 5/105 (4.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. ATPase required during meiosis for the formation of crossover recombination intermediates (By similarity). Binds DNA: preferentially binds to single-stranded DNA and DNA branched structures (PubMed:29742103). Does not show nuclease activity in vitro, but shows ATPase activity, which is stimulated by the presence of single-stranded DNA (PubMed:29742103). Plays a key role in homologous recombination and crossing-over in meiotic prophase ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VXU9 | HPA portal (live; not the pinned values)

SI | Unsafe / Protein

ENSG00000090402

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. duodenum / enterocytes - Microvilli: High (Enhanced); small intestine / enterocytes - Microvilli: High (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (validation): 123/207 (59.4%); Colon Adenocarcinoma (validation): 284/486 (58.4%); Stomach Adenocarcinoma (TCGA): 96/346 (27.7%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Bifunctional enzyme with both sucrase and isomaltase activities involved in breakdown of dietary starch oligosaccharides in small intestine. The isomaltase domain hydrolazes alpha-1,6-glycosidic linkages in isomaltose. The sucrase domain cleaves the alpha-1,2-glycosidic linkages in sucrose to form glucose and fructose, and contributes to the cleavage of the alpha-1,4-glycosidic linkage in maltose to form two glucose monosaccharides

Curated tissue note. Expressed in the poorly differentiated crypt cells of the small intestine as well as in the mature villous cells. Expressed at very low levels in the colon

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P14410 | HPA portal (live; not the pinned values)

SIAH1 | Unsafe / Protein

ENSG00000196470

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Enhanced); adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: High (Enhanced); caudate / neuronal cells: Low (Enhanced); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); ovarian cancer: 12/12 (100.0%)

Biology. E3 ubiquitin-protein ligase that mediates ubiquitination and subsequent proteasomal degradation of target proteins (PubMed:14506261, PubMed:14645235, PubMed:14654780, PubMed:15064394, PubMed:16085652, PubMed:19224863, PubMed:20508617, PubMed:22483617, PubMed:28546513, PubMed:32430360, PubMed:33591310, PubMed:9334332, PubMed:9858595, PubMed:41062835). E3 ubiquitin ligases accept ubiquitin from an E2 ubiquitin-conjugating enzyme in the ... [full text in table]

Curated tissue note. Widely expressed at a low level. Down-regulated in advanced hepatocellular carcinomas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IUQ4 | HPA portal (live; not the pinned values)

SIGLEC6 | Unsafe / Protein

ENSG00000105492

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / mucosal lymphoid cells: Medium (Enhanced)

Cancer RNA. Thyroid Carcinoma (TCGA): 117/495 (23.6%); Breast Invasive Carcinoma (validation): 8/50 (16.0%); Bladder Urothelial Carcinoma (TCGA): 17/169 (10.1%)

Cancer IHC. breast cancer: 1/12 (8.3%); cervical cancer: 0/10 (0.0%)

Biology. Putative adhesion molecule that mediates sialic-acid dependent binding to cells. Binds to alpha-2,6-linked sialic acid. The sialic acid recognition site may be masked by cis interactions with sialic acids on the same cell surface

Curated tissue note. Expressed at high levels in placenta (cyto- and syncytiotrophoblastic cells) and at lower levels in spleen, peripheral blood leukocytes (predominantly B-cells) and small intestine

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43699 | HPA portal (live; not the pinned values)

SIM1 | Unsafe / RNA

ENSG00000112246

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 9.5 nTPM (limit 2); skeletal muscle: 3.4 nTPM (limit 2); kidney: 9.5 nTPM (baseline 2); skeletal muscle: 3.4 nTPM (baseline 2)

Cancer RNA. Kidney Chromophobe (TCGA): 43/64 (67.2%); Kidney Renal Papillary Cell Carcinoma (TCGA): 64/282 (22.7%); Kidney Renal Clear Cell Carcinoma (validation): 10/100 (10.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcriptional factor that may have pleiotropic effects during embryogenesis and in the adult

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P81133 | HPA portal (live; not the pinned values)

SIRPD | Unsafe / Protein

ENSG00000125900

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / cells in tubules: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 5/133 (3.8%); Kidney Chromophobe (TCGA): 1/64 (1.6%); Kidney Renal Clear Cell Carcinoma (validation): 1/100 (1.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H106 | HPA portal (live; not the pinned values)

SIX6 | Unsafe / Protein

ENSG00000184302

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. pituitary gland / cells in anterior : Medium (Approved); retina / inner nuclear layer: Medium (Approved); retina / photoreceptor cells: Low (Approved)

Cancer RNA. Kidney Chromophobe (TCGA): 7/64 (10.9%); Glioblastoma Multiforme (validation): 5/58 (8.6%); Glioblastoma Multiforme (TCGA): 11/141 (7.8%)

Cancer IHC. ovarian cancer: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. May be involved in eye development

Curated tissue note. Expressed in the developing and adult retina. Also expressed in the hypothalamic and the pituitary regions

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95475 | HPA portal (live; not the pinned values)

SKA1 | Unsafe / Protein

ENSG00000154839

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 123/133 (92.5%); Colon Adenocarcinoma (validation): 449/486 (92.4%); Glioblastoma Multiforme (validation): 53/58 (91.4%)

Cancer IHC. breast cancer: 12/12 (100.0%); melanoma: 11/12 (91.7%)

Biology. Component of the SKA complex, a microtubule plus end-binding complex of the outer kinetochore that stabilizes spindle microtubule-kinetochore attachments, promotes alignment of chromosomes at the mitotic spindle equator (chromosome congression) and assists suppression of the spindle assembly checkpoint (PubMed:17093495, PubMed:19289083, PubMed:22371557, PubMed:22483620, PubMed:23085020, PubMed:26981768, PubMed:27697923, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96BD8 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

SKA3 | Unsafe / RNA

ENSG00000165480

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 4.7 nTPM (limit 4); thymus: 11.8 nTPM (limit 10)

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Colon Adenocarcinoma (validation): 459/486 (94.4%); Rectum Adenocarcinoma (validation): 190/207 (91.8%)

Cancer IHC. testis cancer: 2/12 (16.7%); endometrial cancer: 1/12 (8.3%)

Biology. Component of the SKA1 complex, a microtubule-binding subcomplex of the outer kinetochore that is essential for proper chromosome segregation (PubMed:19289083, PubMed:19360002, PubMed:23085020). The SKA1 complex is a direct component of the kinetochore-microtubule interface and directly associates with microtubules as oligomeric assemblies (PubMed:19289083, PubMed:19360002). The complex facilitates the processive movement of ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IX90 | HPA portal (live; not the pinned values)

SKIL | Unsafe / Protein

ENSG00000136603

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebral cortex / endothelial cells: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); skin cancer: 12/12 (100.0%)

Biology. May have regulatory role in cell division or differentiation in response to extracellular signals

Curated tissue note. Isoform SNON and isoform SNOA are widely expressed. Highest expression is found in skeletal muscle, followed by placenta and lung. Lowest expression in heart, brain and pancreas. Isoform SNOI expression is ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P12757 | HPA portal (live; not the pinned values)

SLAIN2 | Unsafe / Protein

ENSG00000109171

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); colon / glandular cells: Low (Approved); duodenum / glandular cells: Low (Approved); esophagus / squamous epithelial cells: Medium (Approved); heart muscle / cardiomyocytes: Medium ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Colon Adenocarcinoma (validation): 486/486 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%)

Cancer IHC. cervical cancer: 3/11 (27.3%); liver cancer: 1/10 (10.0%)

Biology. Binds to the plus end of microtubules and regulates microtubule dynamics and microtubule organization. Promotes cytoplasmic microtubule nucleation and elongation. Required for normal structure of the microtubule cytoskeleton during interphase

Curated tissue note. Widely expressed with highest levels in adult liver, testis and ovary, and lowest levels in adult pancreas and spleen and in fetal brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P270 | HPA portal (live; not the pinned values)

SLC10A7 | Unsafe / RNA

ENSG00000120519

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.4 nTPM (limit 2); bone marrow: 5.9 nTPM (limit 2); cerebral cortex: 2.5 nTPM (limit 2); colon: 6.7 nTPM (limit 6); duodenum: 4.5 nTPM (limit 2); heart muscle: 2.8 nTPM (limit 2); kidney: 3.6 nTPM (limit 2); liver: 7 nTPM (limit 2); lung: 5.7 nTPM (limit 2); midbrain: 2.3 nTPM ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. liver cancer: 9/12 (75.0%); pancreatic cancer: 4/10 (40.0%)

Biology. Involved in teeth and skeletal development. Has an essential role in the biosynthesis and trafficking of glycosaminoglycans and glycoproteins, to produce a proper functioning extracellular matrix. Required for extracellular matrix mineralization (PubMed:29878199, PubMed:30082715). Also involved in the regulation of cellular calcium homeostasis (PubMed:30082715, PubMed:31191616). Does not show transport activity towards bile acids or ... [full text in table]

Curated tissue note. Widely expressed (PubMed:15932064, PubMed:17628207). Expressed at high levels in liver and at lower levels in prostate, placenta, kidney, heart, lung, thymus and spleen (PubMed:15932064, PubMed:17628207). ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q0GE19 | HPA portal (live; not the pinned values)

SLC12A6 | Unsafe / Protein

ENSG00000140199

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebral cortex / glial cells: Low (Uncertain); cerebral cortex / neuronal cells: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. prostate cancer: 12/12 (100.0%); testis cancer: 8/11 (72.7%)

Biology. Mediates electroneutral potassium-chloride cotransport when activated by cell swelling (PubMed:10600773, PubMed:11551954, PubMed:16048901, PubMed:18566107, PubMed:19665974, PubMed:21628467, PubMed:27485015). May contribute to cell volume homeostasis in single cells (PubMed:16048901, PubMed:27485015) Mediates electroneutral potassium-chloride cotransport when activated by cell swelling (PubMed:16048901, PubMed:33199848, ... [full text in table]

Curated tissue note. Expressed in brain (at protein level) (PubMed:21628467). Highly expressed in heart, brain and kidney. Detected at lower levels in skeletal muscle, placenta, lung and pancreas (PubMed:10600773). Detected in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UHW9 | HPA portal (live; not the pinned values)

SLC16A7 | Unsafe / Protein

ENSG00000118596

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / endothelial cells: Medium (Approved); ... [full text in table]

Cancer RNA. Kidney Chromophobe (TCGA): 62/64 (96.9%); Pancreatic Adenocarcinoma (validation): 74/80 (92.5%); Glioblastoma Multiforme (validation): 46/58 (79.3%)

Cancer IHC. breast cancer: 4/10 (40.0%); colorectal cancer: 4/10 (40.0%)

Biology. Proton-coupled monocarboxylate symporter. Catalyzes the rapid transport across the plasma membrane of monocarboxylates such as L-lactate, pyruvate and ketone bodies, acetoacetate, beta-hydroxybutyrate and acetate (PubMed:32415067, PubMed:9786900). Dimerization is functionally required and both subunits work cooperatively in transporting substrate (PubMed:32415067)

Curated tissue note. Detected in heart and in blood lymphocytes and monocytes (at protein level) (PubMed:15505343). High expression in testis, moderate to low in spleen, heart, kidney, pancreas, skeletal muscle, brain and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O60669 | HPA portal (live; not the pinned values)

SLC1A6 | Unsafe / Protein

ENSG00000105143

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Medium (Enhanced); cerebellum / Purkinje cells - cytoplasm/membrane: Medium (Enhanced); cerebellum / Purkinje cells - dendrites: High (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); colon / peripheral nerve/ganglion: Low (Enhanced); lung / macrophages: Low ... [full text in table]

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 69/169 (40.8%); Lung Squamous Cell Carcinoma (TCGA): 96/489 (19.6%); Lung Squamous Cell Carcinoma (validation): 12/68 (17.6%)

Cancer IHC. ovarian cancer: 6/12 (50.0%); urothelial cancer: 3/12 (25.0%)

Biology. Sodium-dependent, high-affinity amino acid transporter that mediates the uptake of L-glutamate and also L-aspartate and D-aspartate (PubMed:26690923, PubMed:7791878). Functions as a symporter that transports one amino acid molecule together with two or three Na(+) ions and one proton, in parallel with the counter-transport of one K(+) ion. Mediates Cl(-) flux that is not coupled to amino acid transport; this avoids the accumulation of ... [full text in table]

Curated tissue note. Brain, mainly in the cerebellum (PubMed:7791878). Expressed densely and selectively in cell bodies of Purkinje cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P48664 | HPA portal (live; not the pinned values)

SLC22A16 | Unsafe / Protein

ENSG00000004809

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 29/176 (16.5%); Ovary Serous Cystadenocarcinoma (TCGA): 8/349 (2.3%); Breast Invasive Carcinoma (validation): 1/50 (2.0%)

Cancer IHC. stomach cancer: 6/9 (66.7%); endometrial cancer: 6/11 (54.5%)

Biology. Facilitative organic cation transporter that mediates the transport of carnitine as well as the polyamine spermidine (PubMed:12089149, PubMed:20037140). Mediates the partially Na(+)-dependent bidirectional transport of carnitine (PubMed:12089149). May mediate L-carnitine secretion from testis epididymal epithelium into the lumen which is involved in the maturation of spermatozoa (PubMed:12089149)

Curated tissue note. Expressed in testis and epididymis (at protein level) (PubMed:12089149, PubMed:12384147, PubMed:15963465). Expressed in endometrium (at protein level); highly expressed during the normal secretory phase, but ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86VW1 | HPA portal (live; not the pinned values)

SLC24A4 | Unsafe / RNA

ENSG00000140090

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.9 nTPM (limit 2); basal ganglia: 3.9 nTPM (limit 2); bone marrow: 3.5 nTPM (limit 2); cerebral cortex: 4.7 nTPM (limit 2); choroid plexus: 44.8 nTPM (limit 2); hypothalamus: 2.1 nTPM (limit 2); midbrain: 5.1 nTPM (limit 2); retina: 12 nTPM (limit 2); spinal cord: 2.3 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 16/58 (27.6%); Skin Cuteneous Melanoma (TCGA): 10/99 (10.1%); Glioblastoma Multiforme (TCGA): 6/141 (4.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Calcium, potassium:sodium antiporter that transports 1 Ca(2+) and 1 K(+) in exchange for 4 Na(+) (PubMed:12379639, PubMed:26631410). Controls the rapid response termination and proper regulation of adaptation in olfactory sensory neurons (OSNs) which subsequently influences how odor information is encoded and perceived (By similarity). May play a role in calcium transport during amelogenesis (PubMed:23375655, PubMed:24621671)

Curated tissue note. Expressed abundantly in all regions of the brain, aorta, lung and thymus (PubMed:12379639). Expressed at lower levels in the stomach and intestine (PubMed:12379639)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NFF2 | HPA portal (live; not the pinned values)

SLC25A31 | Unsafe / Protein

ENSG00000151475

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Uncertain); colon / glandular cells: Low (Uncertain); duodenum / glandular cells: Medium (Uncertain); heart muscle / cardiomyocytes: Low (Uncertain); kidney / cells in tubules: Medium (Uncertain); nasopharynx / respiratory epithelial cells: Low ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 5/133 (3.8%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. glioma: 2/12 (16.7%); melanoma: 1/10 (10.0%)

Biology. ADP:ATP antiporter that mediates import of ADP into the mitochondrial matrix for ATP synthesis, and export of ATP out to fuel the cell (By similarity) (PubMed:15670820). Cycles between the cytoplasmic-open state (c-state) and the matrix-open state (m-state): operates by the alternating access mechanism with a single substrate-binding site intermittently exposed to either the cytosolic (c-state) or matrix (m-state) side of the inner ... [full text in table]

Curated tissue note. Expressed in brain, liver, sperm and testis (PubMed:15670820, PubMed:17137571). In testis, expressed at higher level in spermatocytes, while it is expressed at lower level in spermatogonial cells ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0C2 | HPA portal (live; not the pinned values)

SLC27A6 | Unsafe / Protein

ENSG00000113396

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 355/495 (71.7%); Breast Invasive Carcinoma (validation): 25/50 (50.0%); Ovary Serous Cystadenocarcinoma (validation): 26/81 (32.1%)

Cancer IHC. liver cancer: 3/11 (27.3%); glioma: 2/12 (16.7%)

Biology. Mediates the import of long-chain fatty acids (LCFA) into the cell by facilitating their transport at the plasma membrane (PubMed:12556534). Also functions as an acyl-CoA ligase catalyzing the ATP-dependent formation of fatty acyl-CoA using LCFA and very-long-chain fatty acids (VLCFA) as substrates (By similarity). Plays a pivotal role in regulating available LCFA substrates from exogenous sources in tissues undergoing high levels of ... [full text in table]

Curated tissue note. Strongly expressed in heart and localizes to cardiac myocytes (PubMed:12556534). Expressed at moderate levels in placenta, testis, and adrenal glands. Expressed at very low levels in kidney, bladder and uterus

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y2P4 | HPA portal (live; not the pinned values)

SLC2A14 | Unsafe / Protein

ENSG00000173262

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Supported)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 108/133 (81.2%); Liver Hepatocellular Carcinoma (TCGA): 49/362 (13.5%); Kidney Renal Clear Cell Carcinoma (TCGA): 54/521 (10.4%)

Cancer IHC. testis cancer: 6/12 (50.0%); ovarian cancer: 2/12 (16.7%)

Biology. Hexose transporter that can mediate the transport of glucose and dehydroascorbate across the cell membrane

Curated tissue note. Mainly expressed in testis (PubMed:12504846, PubMed:27460888). Also expressed in small intestine, liver and kidney (PubMed:27460888)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TDB8 | HPA portal (live; not the pinned values)

SLC30A3 | Unsafe / Protein

ENSG00000115194

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / neuronal cells: Low (Enhanced); cerebral cortex / neuropil: Low (Enhanced)

Cancer RNA. Thyroid Carcinoma (TCGA): 82/495 (16.6%); Testicular Germ Cell Tumor (TCGA): 12/133 (9.0%); Liver Hepatocellular Carcinoma (validation): 19/231 (8.2%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Probable proton-coupled zinc ion antiporter mediating the import of zinc from cytoplasm into synaptic vesicles and participating to cellular zinc ion homeostasis in the brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q99726 | HPA portal (live; not the pinned values)

SLC35A1 | Unsafe / RNA

ENSG00000164414

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 15.8 nTPM (limit 10); adrenal gland: 23.2 nTPM (limit 2); amygdala: 17.4 nTPM (limit 2); basal ganglia: 18.9 nTPM (limit 2); bone marrow: 11.1 nTPM (limit 2); cerebellum: 30.8 nTPM (limit 2); cerebral cortex: 21.4 nTPM (limit 2); choroid plexus: 34.8 nTPM (limit 2); colon: 38.3 nTPM ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transports CMP-sialic acid from the cytosol into the Golgi apparatus, functioning as an antiporter that exchanges CMP-sialic acid for CMP (PubMed:12682060, PubMed:15576474, PubMed:23873973). Binds both CMP-sialic acid and free CMP, but has higher affinity for free CMP (By similarity). Also able to exchange CMP-sialic acid for AMP and UMP (PubMed:12682060). Also mediates the transport of CDP-ribitol (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P78382 | HPA portal (live; not the pinned values)

SLC38A9 | Unsafe / Protein

ENSG00000177058

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); colon / glandular cells: Low (Uncertain); duodenum / glandular cells: High (Uncertain); esophagus / squamous epithelial cells: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. liver cancer: 8/12 (66.7%); stomach cancer: 5/12 (41.7%)

Biology. Lysosomal amino acid transporter involved in the activation of mTORC1 in response to amino acid levels (PubMed:25561175, PubMed:25567906, PubMed:29053970). Probably acts as an amino acid sensor of the Rag GTPases and Ragulator complexes, 2 complexes involved in amino acid sensing and activation of mTORC1, a signaling complex promoting cell growth in response to growth factors, energy levels, and amino acids (PubMed:25567906, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NBW4 | HPA portal (live; not the pinned values)

SLC44A5 | Unsafe / RNA

ENSG00000137968

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 3.2 nTPM (limit 2); esophagus: 6.4 nTPM (limit 6); heart muscle: 2.4 nTPM (limit 2); hippocampal formation: 2.3 nTPM (limit 2); hypothalamus: 2.4 nTPM (limit 2); pituitary gland: 3.1 nTPM (limit 2); retina: 2.4 nTPM (limit 2); skin: 10.4 nTPM (limit 10); cerebral cortex: 3.2 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 41/58 (70.7%); Lung Squamous Cell Carcinoma (validation): 45/68 (66.2%); Lung Adenocarcinoma (validation): 62/105 (59.0%)

Cancer IHC. urothelial cancer: 3/10 (30.0%); lung cancer: 2/11 (18.2%)

Biology. Choline/H+ antiporter

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCS7 | HPA portal (live; not the pinned values)

SLC4A5 | Unsafe / Protein

ENSG00000188687

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Uncertain); cerebellum / Purkinje cells: Low (Uncertain); colon / glandular cells: Medium (Uncertain); duodenum / glandular cells: Medium (Uncertain); esophagus / squamous epithelial cells: Low (Uncertain); kidney / cells in tubules: Medium (Uncertain); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 47/50 (94.0%); Thyroid Carcinoma (TCGA): 444/495 (89.7%); Kidney Chromophobe (TCGA): 46/64 (71.9%)

Cancer IHC. breast cancer: 9/11 (81.8%); prostate cancer: 9/12 (75.0%)

Biology. Mediates sodium- and bicarbonate-dependent electrogenic sodium bicarbonate cotransport, with a Na(+):HCO3(-) stoichiometry varying from 1:2 to 1:3

Curated tissue note. Highest expression observed in liver, spleen and testis; moderate expression in the choroid plexus, hippocampus, cerebrum and cerebellum of brain, and in kidney cortex and kidney medulla. Also observed in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BY07 | HPA portal (live; not the pinned values)

SLC52A3 | Unsafe / Protein

ENSG00000101276

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); duodenum / glandular cells: Medium (Approved); esophagus / squamous epithelial cells: Low (Approved); kidney ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%); Rectum Adenocarcinoma (TCGA): 86/88 (97.7%)

Cancer IHC. colorectal cancer: 8/12 (66.7%); melanoma: 5/11 (45.5%)

Biology. Plasma membrane transporter mediating the uptake by cells of the water soluble vitamin B2/riboflavin that plays a key role in biochemical oxidation-reduction reactions of the carbohydrate, lipid, and amino acid metabolism (PubMed:20463145, PubMed:22273710, PubMed:24264046, PubMed:27702554). Humans are unable to synthesize vitamin B2/riboflavin and must obtain it via intestinal absorption (PubMed:20463145)

Curated tissue note. Predominantly expressed in testis. Highly expressed in small intestine and prostate

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NQ40 | HPA portal (live; not the pinned values)

SLC6A16 | Unsafe / RNA

ENSG00000063127

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 3.8 nTPM (limit 2); choroid plexus: 15.5 nTPM (limit 2); heart muscle: 2.4 nTPM (limit 2); kidney: 8.3 nTPM (limit 2); liver: 9.7 nTPM (limit 2); lung: 4.8 nTPM (limit 2); lymph node: 6.7 nTPM (limit 6); pancreas: 6.7 nTPM (limit 2); retina: 4.8 nTPM (limit 2); small intestine: 3.6 ... [full text in table]

Cancer RNA. Kidney Chromophobe (TCGA): 22/64 (34.4%); Kidney Renal Papillary Cell Carcinoma (TCGA): 92/282 (32.6%); Liver Hepatocellular Carcinoma (validation): 71/231 (30.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Highly expressed in peripheral tissues, particularly in testis, pancreas, and prostate

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9GZN6 | HPA portal (live; not the pinned values)

SLC6A2 | Unsafe / Protein

ENSG00000103546

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebellum / processes in granular layer: High (Enhanced); cerebellum / processes in molecular layer: High (Enhanced); cerebellum / processes in white matter: High (Enhanced); cerebral cortex / neuronal projections: High (Enhanced); cerebral cortex / neuropil: Medium (Enhanced)

Cancer RNA. Kidney Chromophobe (TCGA): 19/64 (29.7%); Head and Neck Squamous Cell Carcinoma (TCGA): 130/492 (26.4%); Bladder Urothelial Carcinoma (TCGA): 28/169 (16.6%)

Cancer IHC. colorectal cancer: 3/10 (30.0%); breast cancer: 1/11 (9.1%)

Biology. Mediates sodium- and chloride-dependent transport of norepinephrine (also known as noradrenaline), the primary signaling neurotransmitter in the autonomic sympathetic nervous system (PubMed:2008212, PubMed:8125921, PubMed:38750358). Is responsible for norepinephrine re-uptake and clearance from the synaptic cleft, thus playing a crucial role in norepinephrine inactivation and homeostasis (By similarity). Can also mediate sodium- and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P23975 | HPA portal (live; not the pinned values)

SLC9B1 | Unsafe / High RNA

ENSG00000164037

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 38.8 nTPM; brain/oligodendrocytes; hpa_single_cell_tissue; vital; 5.896 TPM; blood/CD4_NAIVE; dice_cd4_naive; vital

Cancer RNA. Glioblastoma Multiforme (validation): 25/58 (43.1%); Kidney Chromophobe (TCGA): 2/64 (3.1%); Skin Cuteneous Melanoma (TCGA): 3/99 (3.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Sperm-specific Na(+)/H(+) exchanger involved in intracellular pH regulation of spermatozoa. Involved in sperm motility and fertility

Curated tissue note. Expressed only in the testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4ZJI4 | HPA portal (live; not the pinned values)

SLC9C1 | Unsafe / High RNA

ENSG00000172139

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 31.6 nTPM; heart muscle/endothelial cells; hpa_single_cell_tissue; vital; 3.703 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 1/283 (0.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Sperm-specific solute carrier involved in intracellular pH regulation of spermatozoa. Required for sperm motility and fertility. Involved in sperm cell hyperactivation, a step needed for sperm motility which is essential late in the preparation of sperm for fertilization. Required for the expression and bicarbonate regulation of the soluble adenylyl cyclase (ADCY10)

Curated tissue note. Sperm

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4G0N8 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

SLC9C2 | Unsafe / Protein

ENSG00000162753

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); parathyroid gland / glandular cells: Low (Enhanced); smooth muscle / smooth muscle cells: Medium (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 10/58 (17.2%); Ovary Serous Cystadenocarcinoma (validation): 5/81 (6.2%); Pancreatic Adenocarcinoma (validation): 2/80 (2.5%)

Cancer IHC. melanoma: 1/10 (10.0%); breast cancer: 1/11 (9.1%)

Biology. Involved in pH regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5TAH2 | HPA portal (live; not the pinned values)

SLCO4C1 | Unsafe / RNA

ENSG00000173930

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 8.5 nTPM (limit 2); duodenum: 2.1 nTPM (limit 2); kidney: 40.1 nTPM (limit 2); liver: 7.3 nTPM (limit 2); lung: 7.6 nTPM (limit 2); bone marrow: 8.5 nTPM (baseline 2); duodenum: 2.1 nTPM (baseline 2); kidney: 40.1 nTPM (baseline 2); liver: 7.3 nTPM (baseline 2); lung: 7.6 nTPM ... [full text in table]

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 95/100 (95.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 258/282 (91.5%); Kidney Renal Clear Cell Carcinoma (TCGA): 457/521 (87.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Mediates the transport of organic anions such as steroids (estrone 3-sulfate, chenodeoxycholate, glycocholate) and thyroid hormones (3,3',5-triiodo-L-thyronine (T3), L-thyroxine (T4)), in the kidney (PubMed:14993604, PubMed:19129463, PubMed:20610891). Capable of transporting cAMP and pharmacological substances such as digoxin, ouabain and methotrexate (PubMed:14993604). Transport is independent of sodium, chloride ion, and ATP ... [full text in table]

Curated tissue note. Predominantly expressed in kidney but also weakly expressed in both fetal liver and kidney

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZQN7 | HPA portal (live; not the pinned values)

SLIT1 | Unsafe / RNA

ENSG00000187122

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 18.9 nTPM (limit 2); basal ganglia: 23 nTPM (limit 2); cerebral cortex: 33.9 nTPM (limit 2); hippocampal formation: 20 nTPM (limit 2); hypothalamus: 14.3 nTPM (limit 2); midbrain: 5.5 nTPM (limit 2); pituitary gland: 13.1 nTPM (limit 2); amygdala: 18.9 nTPM (baseline 2); basal ganglia: 23 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 106/141 (75.2%); Thyroid Carcinoma (TCGA): 262/495 (52.9%); Prostate Adenocarcinoma (TCGA): 103/480 (21.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Thought to act as molecular guidance cue in cellular migration, and function appears to be mediated by interaction with roundabout homolog receptors. During neural development involved in axonal navigation at the ventral midline of the neural tube and projection of axons to different regions (By similarity). SLIT1 and SLIT2 together seem to be essential for midline guidance in the forebrain by acting as repulsive signal preventing ... [full text in table]

Curated tissue note. Predominantly expressed in adult forebrain. Expressed in fetal brain, lung and kidney

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75093 | HPA portal (live; not the pinned values)

SLX4IP | Unsafe / Protein

ENSG00000149346

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 472/486 (97.1%); Rectum Adenocarcinoma (validation): 199/207 (96.1%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); liver cancer: 11/11 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VYV7 | HPA portal (live; not the pinned values)

SMCP | Unsafe / High RNA

ENSG00000163206

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 45.3 nTPM; testis/macrophages; hpa_single_cell_tissue; vital; 0 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 6/133 (4.5%); Head and Neck Squamous Cell Carcinoma (TCGA): 17/492 (3.5%); Glioblastoma Multiforme (validation): 2/58 (3.4%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Involved in sperm motility. Its absence is associated with genetic background dependent male infertility. Infertility may be due to reduced sperm motility in the female reproductive tract and inability to penetrate the oocyte zona pellucida (By similarity)

Curated tissue note. Testis. Is selectively expressed in the spermatids of seminiferous tubules

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P49901 | HPA portal (live; not the pinned values)

SMIM36 | Unsafe / RNA

ENSG00000261873

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 10.5 nTPM (limit 2); retina: 10.5 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 4/176 (2.3%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 2/283 (0.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0A1B0GVT2 | HPA portal (live; not the pinned values)

SMKR1 | Unsafe / RNA

ENSG00000240204

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 5 nTPM (limit 2); basal ganglia: 10 nTPM (limit 2); cerebral cortex: 6.6 nTPM (limit 2); hippocampal formation: 4.9 nTPM (limit 2); hypothalamus: 4.7 nTPM (limit 2); midbrain: 2.2 nTPM (limit 2); pancreas: 2.2 nTPM (limit 2); parathyroid gland: 6.2 nTPM (limit 6); pituitary gland: 5.9 ... [full text in table]

Cancer RNA. Ovary Serous Cystadenocarcinoma (validation): 57/81 (70.4%); Ovary Serous Cystadenocarcinoma (TCGA): 234/349 (67.0%); Glioblastoma Multiforme (validation): 37/58 (63.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt H3BMG3 | HPA portal (live; not the pinned values)

SNAP91 | Unsafe / Protein

ENSG00000065609

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / granular cells - cytoplasm/membrane: Medium (Enhanced); cerebellum / molecular layer - neuropil: High (Enhanced); cerebellum / processes in granular layer: High (Enhanced); cerebellum / Purkinje cells - ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 35/58 (60.3%); Glioblastoma Multiforme (TCGA): 63/141 (44.7%); Prostate Adenocarcinoma (TCGA): 193/480 (40.2%)

Cancer IHC. renal cancer: 4/11 (36.4%); glioma: 2/9 (22.2%)

Biology. Adaptins are components of the adapter complexes which link clathrin to receptors in coated vesicles. Clathrin-associated protein complexes are believed to interact with the cytoplasmic tails of membrane proteins, leading to their selection and concentration. Binding of AP180 to clathrin triskelia induces their assembly into 60-70 nm coats (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O60641 | HPA portal (live; not the pinned values)

SNAPC3 | Unsafe / RNA

ENSG00000164975

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 7.9 nTPM (limit 2); amygdala: 6.3 nTPM (limit 2); basal ganglia: 6 nTPM (limit 2); cerebellum: 10.2 nTPM (limit 2); cerebral cortex: 6.8 nTPM (limit 2); choroid plexus: 6.1 nTPM (limit 2); duodenum: 3.9 nTPM (limit 2); esophagus: 6.9 nTPM (limit 6); heart muscle: 11.6 nTPM (limit 2); ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 50/50 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Part of the SNAPc complex required for the transcription of both RNA polymerase II and III small-nuclear RNA genes. Binds to the proximal sequence element (PSE), a non-TATA-box basal promoter element common to these 2 types of genes. Recruits TBP and BRF2 to the U6 snRNA TATA box

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92966 | HPA portal (live; not the pinned values)

SNAPC5 | Unsafe / Protein

ENSG00000174446

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. prostate cancer: 9/10 (90.0%); melanoma: 9/12 (75.0%)

Biology. Part of the SNAPc complex required for the transcription of both RNA polymerase II and III small-nuclear RNA genes. Binds to the proximal sequence element (PSE), a non-TATA-box basal promoter element common to these 2 types of genes. Recruits TBP and BRF2 to the U6 snRNA TATA box

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75971 | HPA portal (live; not the pinned values)

SNN | Unsafe / Protein

ENSG00000184602

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Kidney Chromophobe (TCGA): 64/64 (100.0%)

Cancer IHC. testis cancer: 12/12 (100.0%); melanoma: 10/10 (100.0%)

Biology. Plays a role in the toxic effects of organotins (PubMed:15269288). Plays a role in endosomal maturation (PubMed:27015288)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75324 | HPA portal (live; not the pinned values)

SNX16 | Unsafe / Protein

ENSG00000104497

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / ciliated cells (cell body): Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Adenocarcinoma (validation): 103/105 (98.1%)

Cancer IHC. liver cancer: 9/12 (75.0%); cervical cancer: 7/12 (58.3%)

Biology. May be involved in several stages of intracellular trafficking. Plays a role in protein transport from early to late endosomes. Plays a role in protein transport to the lysosome. Promotes degradation of EGFR after EGF signaling. Plays a role in intracellular transport of vesicular stomatitis virus nucleocapsids from the endosome to the cytoplasm

Curated tissue note. Detected in placenta, lung, liver,heart and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P57768 | HPA portal (live; not the pinned values)

SNX19 | Unsafe / Protein

ENSG00000120451

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); cerebral cortex / glial cells: Medium (Uncertain); cerebral cortex / neuropil: Low (Uncertain); colon / peripheral nerve/ganglion: Low (Uncertain); duodenum / glandular cells: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. endometrial cancer: 9/12 (75.0%); colorectal cancer: 6/11 (54.5%)

Biology. Plays a role in intracellular vesicle trafficking and exocytosis (PubMed:24843546). May play a role in maintaining insulin-containing dense core vesicles in pancreatic beta-cells and in preventing their degradation. May play a role in insulin secretion (PubMed:24843546). Interacts with membranes containing phosphatidylinositol 3-phosphate (PtdIns(3P)) (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92543 | HPA portal (live; not the pinned values)

SOHLH1 | Unsafe / RNA

ENSG00000165643

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 8.5 nTPM (limit 2); basal ganglia: 5 nTPM (limit 2); cerebellum: 3.6 nTPM (limit 2); cerebral cortex: 41.6 nTPM (limit 2); hippocampal formation: 18.8 nTPM (limit 2); hypothalamus: 2.8 nTPM (limit 2); midbrain: 2.7 nTPM (limit 2); pituitary gland: 3.1 nTPM (limit 2); amygdala: 8.5 nTPM ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 145/489 (29.7%); Head and Neck Squamous Cell Carcinoma (TCGA): 60/492 (12.2%); Lung Squamous Cell Carcinoma (validation): 8/68 (11.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcription regulator of both male and female germline differentiation. Suppresses genes involved in spermatogonial stem cells maintenance, and induces genes important for spermatogonial differentiation. Coordinates oocyte differentiation without affecting meiosis I (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JUK2 | HPA portal (live; not the pinned values)

SOS2 | Unsafe / Protein

ENSG00000100485

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); melanoma: 12/12 (100.0%)

Biology. Acts as guanine nucleotide exchange factor (GEF) for RAS proteins. Catalyzes the GDP-to-GTP exchange, resulting in an increase of the active GTP-bound form of HRAS (PubMed:20639119). Acts as a key modulator of PI3K-AKT signaling

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q07890 | HPA portal (live; not the pinned values)

SOX1 | Unsafe / RNA

ENSG00000182968

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 4 nTPM (limit 2); basal ganglia: 3.9 nTPM (limit 2); cerebral cortex: 3.4 nTPM (limit 2); hippocampal formation: 2.4 nTPM (limit 2); hypothalamus: 5.4 nTPM (limit 2); amygdala: 4 nTPM (baseline 2); basal ganglia: 3.9 nTPM (baseline 2); cerebral cortex: 3.4 nTPM (baseline 2); hippocampal ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 18/58 (31.0%); Glioblastoma Multiforme (TCGA): 23/141 (16.3%); Rectum Adenocarcinoma (TCGA): 9/88 (10.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcriptional activator. May function as a switch in neuronal development. Keeps neural cells undifferentiated by counteracting the activity of proneural proteins and suppresses neuronal differentiation (By similarity)

Curated tissue note. Mainly expressed in the developing central nervous system

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00570 | HPA portal (live; not the pinned values)

SOX3 | Unsafe / RNA

ENSG00000134595

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. hypothalamus: 3.1 nTPM (limit 2); hypothalamus: 3.1 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (TCGA): 30/141 (21.3%); Glioblastoma Multiforme (validation): 10/58 (17.2%); Breast Invasive Carcinoma (validation): 5/50 (10.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcription factor required during the formation of the hypothalamo-pituitary axis. May function as a switch in neuronal development. Keeps neural cells undifferentiated by counteracting the activity of proneural proteins and suppresses neuronal differentiation. Required also within the pharyngeal epithelia for craniofacial morphogenesis. Controls a genetic switch in male development. Is necessary for initiating male sex ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P41225 | HPA portal (live; not the pinned values)

SOX6 | Unsafe / Protein

ENSG00000110693

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / cells in granular layer: Low (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Kidney Renal Clear Cell Carcinoma (validation): 64/100 (64.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 179/282 (63.5%)

Cancer IHC. glioma: 7/11 (63.6%); melanoma: 3/12 (25.0%)

Biology. Transcription factor that plays a key role in several developmental processes, including neurogenesis, chondrocytes differentiation and cartilage formation (Probable). Specifically binds the 5'-AACAAT-3' DNA motif present in enhancers and super-enhancers and promotes expression of genes important for chondrogenesis. Required for overt chondrogenesis when condensed prechondrocytes differentiate into early stage chondrocytes: SOX5 and ... [full text in table]

Curated tissue note. Expressed in a wide variety of tissues, most abundantly in skeletal musclen

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P35712 | HPA portal (live; not the pinned values)

SPA17 | Unsafe / Protein

ENSG00000064199

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. 345.8 nTPM; lung/ciliated cells; hpa_single_cell_tissue; vital; 2.327 TPM; blood/M2; dice_m2; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 50/50 (100.0%); Pancreatic Adenocarcinoma (validation): 75/80 (93.8%); Glioblastoma Multiforme (validation): 53/58 (91.4%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/9 (0.0%)

Biology. Sperm surface zona pellucida binding protein. Helps to bind spermatozoa to the zona pellucida with high affinity. Might function in binding zona pellucida and carbohydrates (By similarity)

Curated tissue note. Testis and sperm specific

Therapeutic evidence. Trial registration only.

UniProt Q15506 | Trial/source record | HPA portal (live; not the pinned values)

SPACA3 | Unsafe / RNA

ENSG00000141316

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pancreas: 20.3 nTPM (limit 12)

Cancer RNA. Rectum Adenocarcinoma (TCGA): 13/88 (14.8%); Colon Adenocarcinoma (TCGA): 17/254 (6.7%); Skin Cuteneous Melanoma (TCGA): 5/99 (5.1%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Sperm surface membrane protein that may be involved in sperm-egg plasma membrane adhesion and fusion during fertilization. It could be a potential receptor for the egg oligosaccharide residue N-acetylglucosamine, which is present in the extracellular matrix over the egg plasma membrane. The processed form has no detectable bacteriolytic activity in vitro

Curated tissue note. The processed form is expressed in sperm (at protein level). Expressed in testis, epididymis and placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IXA5 | HPA portal (live; not the pinned values)

SPACA9 | Unsafe / Protein

ENSG00000165698

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (ciliary rootlets): High (Enhanced); nasopharynx / ciliated cells (tip of cilia): High (Enhanced)

Cancer RNA. Glioblastoma Multiforme (TCGA): 139/141 (98.6%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Thyroid Carcinoma (TCGA): 485/495 (98.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) of multiciliated respiratory cells and the distal singlet microtubules of monoflagellated spermatozoa (PubMed:36191189). Forms an extensive interaction network cross-linking the lumen of axonemal doublet microtubules (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96E40 | HPA portal (live; not the pinned values)

SPACDR | Unsafe / RNA

ENSG00000185955

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 7.3 nTPM (limit 2); basal ganglia: 7.6 nTPM (limit 2); hippocampal formation: 8.1 nTPM (limit 2); hypothalamus: 6.8 nTPM (limit 2); midbrain: 9.1 nTPM (limit 2); pituitary gland: 2.8 nTPM (limit 2); spinal cord: 8.8 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (TCGA): 126/141 (89.4%); Glioblastoma Multiforme (validation): 20/58 (34.5%); Uterine Corpus Endometrial Carcinoma (TCGA): 11/176 (6.2%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May play a role in acrosome formation and nucleus shaping during spermiogenesis

Curated tissue note. Expressed in sperm (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZ16 | HPA portal (live; not the pinned values)

SPAG1 | Unsafe / Protein

ENSG00000104450

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); nasopharynx / ciliated cells (cell body): Medium (Enhanced); rectum / glandular cells: Low (Enhanced); skeletal muscle / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 205/207 (99.0%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 49/50 (98.0%)

Cancer IHC. glioma: 1/11 (9.1%); cervical cancer: 1/12 (8.3%)

Biology. May play a role in the cytoplasmic assembly of the ciliary dynein arms (By similarity). May play a role in fertilization. Binds GTP and has GTPase activity

Curated tissue note. Present in most tissues, including lung, with the strongest expression in brain, colon, kidney, and testis. In sperm and testis, detected in particular in pachytene primary spermatocytes. Up-regulated in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q07617 | HPA portal (live; not the pinned values)

SPAG6 | Unsafe / Protein

ENSG00000077327

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); caudate / glial cells: High (Enhanced); lung / macrophages: Medium (Enhanced); nasopharynx / ciliated cells (cell body): Medium (Enhanced); nasopharynx / ciliated cells (cilia ... [full text in table]

Cancer RNA. Kidney Renal Papillary Cell Carcinoma (TCGA): 100/282 (35.5%); Breast Invasive Carcinoma (validation): 12/50 (24.0%); Lung Adenocarcinoma (validation): 24/105 (22.9%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Important for structural integrity of the central apparatus in the sperm tail and for flagellar motility

Curated tissue note. Highly expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75602 | HPA portal (live; not the pinned values)

SPAG9 | Unsafe / Protein

ENSG00000008294

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. 480.1 nTPM; adipose tissue/macrophages; hpa_single_cell_tissue; vital; 207.5 TPM; blood/CD4_STIM; dice_cd4_stim; vital

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. testis cancer: 11/11 (100.0%); colorectal cancer: 10/12 (83.3%)

Biology. The JNK-interacting protein (JIP) group of scaffold proteins selectively mediates JNK signaling by aggregating specific components of the MAPK cascade to form a functional JNK signaling module (PubMed:14743216). Regulates lysosomal positioning by acting as an adapter protein which links PIP4P1-positive lysosomes to the dynein-dynactin complex (PubMed:29146937). Assists PIKFYVE selective functionality in microtubule-based ... [full text in table]

Curated tissue note. Expressed only in testis on the round spermatids of stage I, II and II. Absent in spermatogonia and spermatocyte Expressed in testis and in acute myeloid leukemia (AML) patients Expressed in testis

Therapeutic evidence. Tumor expression and immunity.

UniProt O60271 | Reviewed source 1 | HPA portal (live; not the pinned values)

SPAST | Unsafe / Protein

ENSG00000021574

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); stomach cancer: 11/11 (100.0%)

Biology. ATP-dependent microtubule severing protein that specifically recognizes and cuts microtubules that are polyglutamylated (PubMed:11809724, PubMed:15716377, PubMed:16219033, PubMed:17389232, PubMed:20530212, PubMed:22637577, PubMed:26875866). Preferentially recognizes and acts on microtubules decorated with short polyglutamate tails: severing activity increases as the number of glutamates per tubulin rises from one to eight, but ... [full text in table]

Curated tissue note. Expressed in brain, heart, kidney, liver, lung, pancreas, placenta and skeletal muscle. The short isoforms may predominate in brain and spinal cord

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UBP0 | HPA portal (live; not the pinned values)

SPATA17 | Unsafe / Protein

ENSG00000162814

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); kidney / cells in tubules: Low (Enhanced); nasopharynx / ciliated cells (cilia axoneme): Medium (Enhanced); nasopharynx / ciliated cells (tip of cilia): Low (Enhanced); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Glioblastoma Multiforme (validation): 22/58 (37.9%); Ovary Serous Cystadenocarcinoma (validation): 20/81 (24.7%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96L03 | HPA portal (live; not the pinned values)

SPATA22 | Unsafe / High RNA

ENSG00000141255

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 47.2 nTPM; brain/oligodendrocytes; hpa_single_cell_tissue; vital; 3.496 TPM; blood/naive CD8 T-cell; hpa_immune_cell_samples; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 87/133 (65.4%); Glioblastoma Multiforme (validation): 3/58 (5.2%); Bladder Urothelial Carcinoma (TCGA): 5/169 (3.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Meiosis-specific protein required for homologous recombination in meiosis I

Curated tissue note. Highly expressed in adult testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NHS9 | HPA portal (live; not the pinned values)

SPATA24 | Unsafe / RNA

ENSG00000170469

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 6.1 nTPM (limit 2); basal ganglia: 5.5 nTPM (limit 2); choroid plexus: 9.7 nTPM (limit 2); heart muscle: 19.9 nTPM (limit 12); hippocampal formation: 5.3 nTPM (limit 2); hypothalamus: 6.1 nTPM (limit 2); kidney: 14.4 nTPM (limit 12); midbrain: 5.9 nTPM (limit 2); pituitary gland: 12.3 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Prostate Adenocarcinoma (TCGA): 453/480 (94.4%); Rectum Adenocarcinoma (validation): 194/207 (93.7%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/9 (0.0%)

Biology. Binds DNA with high affinity but does not bind to TATA boxes. Synergises with GMNN and TBP in activation of TATA box-containing promoters and with GMNN and TBPL1 in activation of the NF1 TATA-less promoter. May play a role in cytoplasm movement and removal during spermiogenesis (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86W54 | HPA portal (live; not the pinned values)

SPATA31G1 | Unsafe / RNA

ENSG00000174038

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.6 nTPM (limit 2); bone marrow: 2.6 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 152/207 (73.4%); Colon Adenocarcinoma (validation): 313/486 (64.4%); Bladder Urothelial Carcinoma (TCGA): 14/169 (8.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Dispensable for normal development and fertility

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VYM1 | HPA portal (live; not the pinned values)

SPATA33 | Unsafe / Protein

ENSG00000167523

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (ciliary rootlets): Low (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); colon / enterocytes: Low (Enhanced); duodenum / paneth ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 202/207 (97.6%); Colon Adenocarcinoma (validation): 463/486 (95.3%); Glioblastoma Multiforme (TCGA): 125/141 (88.7%)

Cancer IHC. breast cancer: 0/9 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Plays an important role in sperm motility and male fertility (By similarity). Required for sperm midpiece flexibility and for the localization of sperm calcineurin to the mitochondria (By similarity). Promotes mitophagy as well as acts as an autophagy mediator in male germline cells (By similarity). Links damaged mitochondria to autophagosomes via its binding to the outer mitochondrial membrane protein VDAC2, as well as to key ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96N06 | HPA portal (live; not the pinned values)

SPATA4 | Unsafe / RNA

ENSG00000150628

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. retina: 2.2 nTPM (limit 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 11/50 (22.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 10/176 (5.7%); Glioblastoma Multiforme (TCGA): 3/141 (2.1%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May play a role in apoptosis regulation

Curated tissue note. Highly expressed in testis, the expression is observed precisely in seminiferous tubules

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEY3 | HPA portal (live; not the pinned values)

SPATA45 | Unsafe / RNA

ENSG00000185523

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pancreas: 2.9 nTPM (limit 2); pancreas: 2.9 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 6/58 (10.3%); Kidney Chromophobe (TCGA): 1/64 (1.6%); Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%)

Cancer IHC. renal cancer: 11/12 (91.7%); urothelial cancer: 8/10 (80.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q537H7 | HPA portal (live; not the pinned values)

SPATA46 | Unsafe / RNA

ENSG00000171722

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.1 nTPM (limit 2); retina: 2.3 nTPM (limit 2); adrenal gland: 3.1 nTPM (baseline 2); retina: 2.3 nTPM (baseline 2)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Breast Invasive Carcinoma (TCGA): 12/1022 (1.2%); Lung Adenocarcinoma (validation): 1/105 (1.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays a role in spermiogenesis and fertilization

Curated tissue note. Testis-specific (PubMed:27488028)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T0L3 | HPA portal (live; not the pinned values)

SPATA6 | Unsafe / RNA

ENSG00000132122

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 7.8 nTPM (limit 2); amygdala: 5.4 nTPM (limit 2); basal ganglia: 5.1 nTPM (limit 2); cerebral cortex: 3.8 nTPM (limit 2); choroid plexus: 6.1 nTPM (limit 2); duodenum: 2.6 nTPM (limit 2); heart muscle: 4.2 nTPM (limit 2); hippocampal formation: 5.7 nTPM (limit 2); hypothalamus: 5.1 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 135/141 (95.7%); Pancreatic Adenocarcinoma (validation): 72/80 (90.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Required for formation of the sperm connecting piece during spermiogenesis. Sperm connecting piece is essential for linking the developing flagellum to the head during late spermiogenesis. May be involved in myosin-based microfilament transport through interaction with myosin subunits

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NWH7 | HPA portal (live; not the pinned values)

SPATA9 | Unsafe / Protein

ENSG00000145757

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); cerebral cortex / glial cells: Low (Approved); cerebral cortex / neuropil: Low (Approved); colon / glandular cells: Low (Approved); colon / peripheral ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 3/50 (6.0%); Glioblastoma Multiforme (validation): 2/58 (3.4%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. glioma: 11/11 (100.0%); colorectal cancer: 5/12 (41.7%)

Biology. May play a role in testicular development/spermatogenesis and may be an important factor in male infertility

Curated tissue note. Highly expressed in testes and pancreas. Low levels found in the heart, lungs, and brain. Very low expression detected in the placenta. No expression seen in skeletal muscle, liver, kidney, thymus, small ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BWV2 | HPA portal (live; not the pinned values)

SPATC1 | Unsafe / RNA

ENSG00000186583

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 8.4 nTPM (limit 2); bone marrow: 8.4 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 2/176 (1.1%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 2/282 (0.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Detected only in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q76KD6 | HPA portal (live; not the pinned values)

SPC25 | Unsafe / Protein

ENSG00000152253

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); colon / glandular cells: Medium (Enhanced); duodenum / glandular cells: Low (Enhanced); esophagus / squamous epithelial cells: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); lymph node / germinal center cells: High (Enhanced); lymph ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 79/80 (98.8%); Glioblastoma Multiforme (validation): 55/58 (94.8%); Breast Invasive Carcinoma (validation): 46/50 (92.0%)

Cancer IHC. cervical cancer: 8/12 (66.7%); testis cancer: 8/12 (66.7%)

Biology. Acts as a component of the essential kinetochore-associated NDC80 complex, which is required for chromosome segregation and spindle checkpoint activity (PubMed:14699129, PubMed:14738735). Required for kinetochore integrity and the organization of stable microtubule binding sites in the outer plate of the kinetochore (PubMed:14699129, PubMed:14738735). The NDC80 complex synergistically enhances the affinity of the SKA1 complex for ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HBM1 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

SPDYA | Unsafe / High RNA

ENSG00000163806

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 37 nTPM; skeletal muscle/macrophages; hpa_single_cell_tissue; vital; 16.88 TPM; blood/MONOCYTES; dice_monocytes; vital

Cancer RNA. Prostate Adenocarcinoma (TCGA): 407/480 (84.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 119/176 (67.6%); Liver Hepatocellular Carcinoma (validation): 143/231 (61.9%)

Cancer IHC. colorectal cancer: 1/10 (10.0%); skin cancer: 1/10 (10.0%)

Biology. Regulates the G1/S phase transition of the cell cycle by binding and activating CDK1 and CDK2 (PubMed:12972555). Contributes to CDK2 activation without promoting CDK2 phosphorylation, by inducing a conformation change of the CDK2 T-loop that obstructs the substrate-binding cleft prior to kinase activation (PubMed:28666995). Mediates cell survival during the DNA damage process through activation of CDK2 (PubMed:12839962)

Curated tissue note. Highly expressed in testis. Expressed at a low level in wide range of tissues including bone marrow, brain, heart, kidney, colon, liver, placenta, spleen, skeletal muscle, salivary gland, thyroid gland, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5MJ70 | HPA portal (live; not the pinned values)

SPDYE17 | Unsafe / Protein

ENSG00000186645

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / cells in tubules: Low (Supported); thyroid gland / glandular cells: Low (Supported)

Cancer RNA. Rectum Adenocarcinoma (validation): 122/207 (58.9%); Colon Adenocarcinoma (validation): 231/486 (47.5%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Acts as a modulator of the nicotinic acetylcholine receptor alpha-7 (CHRNA7) activity

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0A494C191 | UniProt P0DTA3 | UniProt P0DUD1 | UniProt P0DUD2 | UniProt P0DUX0 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

SPECC1 | Unsafe / Protein

ENSG00000128487

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Enhanced); adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / ciliated cells (cell body): High (Enhanced); bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. glioma: 11/12 (91.7%); stomach cancer: 11/12 (91.7%)

Biology. Highly expressed in testis. Barely detectable in other tissues. Also highly expressed in some cancer cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5M775 | HPA portal (live; not the pinned values)

SPEF2 | Unsafe / Protein

ENSG00000152582

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cilia axoneme): High (Enhanced); nasopharynx / ciliated cells (tip of cilia): High (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 48/50 (96.0%); Kidney Renal Clear Cell Carcinoma (validation): 74/100 (74.0%); Ovary Serous Cystadenocarcinoma (validation): 41/81 (50.6%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Required for correct axoneme development in spermatozoa. Important for normal development of the manchette and sperm head morphology. Essential for male fertility. Plays a role in localization of the intraflagellar transport protein IFT20 to the manchette, suggesting function as an adapter for dynein-mediated protein transport during spermatogenesis (PubMed:31048344, PubMed:31151990, PubMed:31278745). Plays a role in bone growth where ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9C093 | HPA portal (live; not the pinned values)

SPINK2 | Unsafe / RNA

ENSG00000128040

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 13.1 nTPM (limit 12); choroid plexus: 9.3 nTPM (limit 2); pituitary gland: 4 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 55/133 (41.4%); Thyroid Carcinoma (TCGA): 166/495 (33.5%); Breast Invasive Carcinoma (validation): 8/50 (16.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. As a strong inhibitor of acrosin, it is required for normal spermiogenesis. It probably hinders premature activation of proacrosin and other proteases, thus preventing the cascade of events leading to spermiogenesis defects (PubMed:28554943). May be involved in the regulation of serine protease-dependent germ cell apoptosis (By similarity). It also inhibits trypsin

Curated tissue note. Expressed in epididymis (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P20155 | HPA portal (live; not the pinned values)

SPINT3 | Unsafe / Protein

ENSG00000101446

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. skin 1 / fibrohistiocytic cells: Medium (Enhanced); skin 1 / lymphocytes: High (Enhanced); skin 2 / fibrohistiocytic cells: Low (Enhanced); skin 2 / lymphocytes: Medium (Enhanced)

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 3/169 (1.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 2/282 (0.7%); Liver Hepatocellular Carcinoma (TCGA): 2/362 (0.6%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P49223 | HPA portal (live; not the pinned values)

SPINT4 | Unsafe / Protein

ENSG00000149651

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); salivary gland / glandular cells: Low (Enhanced); small intestine / glandular cells: Low (Enhanced); spleen / cells in red pulp: Low (Enhanced); spleen / cells in white pulp: Low (Enhanced)

Cancer RNA. Rectum Adenocarcinoma (validation): 6/207 (2.9%); Colon Adenocarcinoma (validation): 5/486 (1.0%); Colon Adenocarcinoma (TCGA): 2/254 (0.8%)

Cancer IHC. liver cancer: 3/9 (33.3%); melanoma: 2/11 (18.2%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6UDR6 | HPA portal (live; not the pinned values)

SPMIP4 | Unsafe / RNA

ENSG00000153790

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 7.6 nTPM (limit 2); basal ganglia: 2.5 nTPM (limit 2); cerebellum: 3.8 nTPM (limit 2); colon: 6.6 nTPM (limit 6); duodenum: 4.3 nTPM (limit 2); heart muscle: 2.1 nTPM (limit 2); kidney: 2.7 nTPM (limit 2); lung: 2.5 nTPM (limit 2); pituitary gland: 2.1 nTPM (limit 2); retina: 2.6 ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 87/133 (65.4%); Rectum Adenocarcinoma (validation): 129/207 (62.3%); Breast Invasive Carcinoma (validation): 31/50 (62.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in flagellum axoneme. May serve to reinforce and thus stabilize the microtubule structure in the sperm flagella

Curated tissue note. Predominantly expressed in the testes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N865 | HPA portal (live; not the pinned values)

SPMIP5 | Unsafe / High RNA

ENSG00000165863

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 29.33 nTPM; blood/basophil; hpa_immune_cell_samples; vital; 30.8 TPM; blood/basophil; hpa_immune_monaco; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 32/50 (64.0%); Breast Invasive Carcinoma (TCGA): 146/1022 (14.3%); Uterine Corpus Endometrial Carcinoma (TCGA): 19/176 (10.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in flagellum axoneme. May serve to reinforce and thus stabilize the microtubule structure in the sperm flagella

Curated tissue note. Expressed in testis (at protein level). Strongly expressed in peritubular cells and Leydig cells and weakly expressed in the cytoplasm of spermatocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WW14 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

SPRED1 | Unsafe / Protein

ENSG00000166068

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. liver cancer: 6/12 (50.0%); glioma: 4/11 (36.4%)

Biology. Tyrosine kinase substrate that inhibits growth-factor-mediated activation of MAP kinase (By similarity). Negatively regulates hematopoiesis of bone marrow (By similarity). Inhibits fibroblast growth factor (FGF)-induced retinal lens fiber differentiation, probably by inhibiting FGF-mediated phosphorylation of ERK1/2 (By similarity). Attenuates actin stress fiber formation via inhibition of TESK1-mediated phosphorylation of cofilin ... [full text in table]

Curated tissue note. Weakly expressed in embryonic cell line HEK293

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z699 | HPA portal (live; not the pinned values)

SPRN | Unsafe / RNA

ENSG00000203772

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4 nTPM (limit 2); amygdala: 9.4 nTPM (limit 2); basal ganglia: 8.7 nTPM (limit 2); cerebellum: 16.7 nTPM (limit 2); cerebral cortex: 25.1 nTPM (limit 2); hippocampal formation: 13.3 nTPM (limit 2); hypothalamus: 6.5 nTPM (limit 2); kidney: 5.3 nTPM (limit 2); liver: 17.3 nTPM (limit ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 42/50 (84.0%); Liver Hepatocellular Carcinoma (validation): 105/231 (45.5%); Uterine Corpus Endometrial Carcinoma (TCGA): 65/176 (36.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Prion-like protein that has PrP(C)-like neuroprotective activity. May act as a modulator for the biological actions of normal and abnormal PrP (By similarity)

Curated tissue note. Mainly expressed in brain. In brain, it is expressed in hippocampus

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5BIV9 | HPA portal (live; not the pinned values)

SPRR4 | Unsafe / Protein

ENSG00000184148

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. esophagus / squamous epithelial cells: Low (Enhanced); oral mucosa / squamous epithelial cells: Low (Enhanced); skin 1 / cells in granular layer: Medium (Enhanced); skin 2 / cells in granular layer: Medium (Enhanced); skin 2 / cells in spinous layer: Low (Enhanced)

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 263/492 (53.5%); Skin Cuteneous Melanoma (TCGA): 19/99 (19.2%); Lung Squamous Cell Carcinoma (TCGA): 70/489 (14.3%)

Cancer IHC. skin cancer: 6/10 (60.0%); cervical cancer: 5/12 (41.7%)

Biology. Cross-linked envelope protein of keratinocytes. Involved in UV-induced cornification

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96PI1 | HPA portal (live; not the pinned values)

SPRTN | Unsafe / Protein

ENSG00000010072

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in molecular layer: Medium (Uncertain); cerebellum / Purkinje cells: High (Uncertain); ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 204/207 (98.6%)

Cancer IHC. melanoma: 1/11 (9.1%); cervical cancer: 1/12 (8.3%)

Biology. DNA-dependent metalloendopeptidase that mediates the proteolytic cleavage of covalent DNA-protein cross-links (DPCs) during DNA synthesis and mitosis, thereby playing a key role in maintaining genomic integrity (PubMed:27852435, PubMed:27871365, PubMed:27871366, PubMed:30893605, PubMed:32649882, PubMed:36608669, PubMed:41610251). DPCs are highly toxic DNA lesions that interfere with essential chromatin transactions, such as replication ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H040 | HPA portal (live; not the pinned values)

SPTA1 | Unsafe / Protein

ENSG00000163554

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: High (Enhanced)

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%); Liver Hepatocellular Carcinoma (validation): 3/231 (1.3%); Liver Hepatocellular Carcinoma (TCGA): 3/362 (0.8%)

Cancer IHC. testis cancer: 9/12 (75.0%); renal cancer: 7/12 (58.3%)

Biology. Spectrin is the major constituent of the cytoskeletal network underlying the erythrocyte plasma membrane. It associates with band 4.1 and actin to form the cytoskeletal superstructure of the erythrocyte plasma membrane

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P02549 | HPA portal (live; not the pinned values)

SRSF12 | Unsafe / RNA

ENSG00000154548

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.7 nTPM (limit 2); basal ganglia: 3.3 nTPM (limit 2); cerebral cortex: 4.2 nTPM (limit 4); hippocampal formation: 3 nTPM (limit 2); hypothalamus: 3.9 nTPM (limit 2); pituitary gland: 2.5 nTPM (limit 2); retina: 2.3 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Testicular Germ Cell Tumor (TCGA): 39/133 (29.3%); Glioblastoma Multiforme (TCGA): 39/141 (27.7%)

Cancer IHC. colorectal cancer: 2/11 (18.2%); cervical cancer: 1/8 (12.5%)

Biology. Splicing factor that seems to antagonize SR proteins in pre-mRNA splicing regulation

Curated tissue note. Expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WXF0 | HPA portal (live; not the pinned values)

SSX2IP | Unsafe / Protein

ENSG00000117155

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / glial cells: Medium (Supported); caudate / neuronal cells: High (Supported); cerebellum / Purkinje cells: Low (Supported); ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 484/486 (99.6%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); endometrial cancer: 11/11 (100.0%)

Biology. Belongs to an adhesion system, which plays a role in the organization of homotypic, interneuronal and heterotypic cell-cell adherens junctions (AJs). May connect the nectin-afadin and E-cadherin-catenin system through alpha-actinin and may be involved in organization of the actin cytoskeleton at AJs through afadin and alpha-actinin (By similarity). Involved in cell movement: localizes at the leading edge of moving cells in response to ... [full text in table]

Curated tissue note. Widely expressed, with the highest expression in brain, intermediate expression in kidney, testis, spinal cord, liver, heart, lung, skeletal muscle, ovary, fetal liver and fetal brain, and little to no ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y2D8 | HPA portal (live; not the pinned values)

STAG3 | Unsafe / Protein

ENSG00000066923

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. heart muscle / cardiomyocytes: Low (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 26/50 (52.0%); Ovary Serous Cystadenocarcinoma (validation): 29/81 (35.8%); Liver Hepatocellular Carcinoma (validation): 63/231 (27.3%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Meiosis specific component of cohesin complex. The cohesin complex is required for the cohesion of sister chromatids after DNA replication. The cohesin complex apparently forms a large proteinaceous ring within which sister chromatids can be trapped. At anaphase, the complex is cleaved and dissociates from chromatin, allowing sister chromatids to segregate. The meiosis-specific cohesin complex probably replaces mitosis specific cohesin ... [full text in table]

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UJ98 | HPA portal (live; not the pinned values)

STAP1 | Unsafe / Protein

ENSG00000035720

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); colon / endocrine cells: Low (Enhanced); colon / enterocytes: Low (Enhanced); colon / goblet cells: Low (Enhanced); colon / mucosal lymphoid cells: High (Enhanced); duodenum / endocrine cells: Medium (Enhanced); duodenum / enterocytes: Medium ... [full text in table]

Cancer RNA. Kidney Chromophobe (TCGA): 58/64 (90.6%); Breast Invasive Carcinoma (validation): 24/50 (48.0%); Lung Adenocarcinoma (validation): 13/105 (12.4%)

Cancer IHC. lymphoma: 6/11 (54.5%); prostate cancer: 2/10 (20.0%)

Biology. In BCR signaling, appears to function as a docking protein acting downstream of TEC and participates in a positive feedback loop by increasing the activity of TEC

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9ULZ2 | HPA portal (live; not the pinned values)

STEAP1B | Unsafe / RNA

ENSG00000105889

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.3 nTPM (limit 2); choroid plexus: 5.5 nTPM (limit 2); hippocampal formation: 2.1 nTPM (limit 2); midbrain: 2.5 nTPM (limit 2); spinal cord: 3.1 nTPM (limit 2); basal ganglia: 2.3 nTPM (baseline 2); choroid plexus: 5.5 nTPM (baseline 2); hippocampal formation: 2.1 nTPM (baseline 2); ... [full text in table]

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 66/99 (66.7%); Lung Squamous Cell Carcinoma (validation): 28/68 (41.2%); Head and Neck Squamous Cell Carcinoma (TCGA): 202/492 (41.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6NZ63 | HPA portal (live; not the pinned values)

STIL | Unsafe / RNA

ENSG00000123473

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 8.8 nTPM (limit 2); duodenum: 2.6 nTPM (limit 2); small intestine: 2.3 nTPM (limit 2); bone marrow: 8.8 nTPM (baseline 2); duodenum: 2.6 nTPM (baseline 2); small intestine: 2.3 nTPM (baseline 2)

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 204/207 (98.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Immediate-early gene. Plays an important role in embryonic development as well as in cellular growth and proliferation; its long-term silencing affects cell survival and cell cycle distribution as well as decreases CDK1 activity correlated with reduced phosphorylation of CDK1. Plays a role as a positive regulator of the sonic hedgehog pathway, acting downstream of PTCH1 (PubMed:16024801, PubMed:9372240). Plays an important role in the ... [full text in table]

Curated tissue note. Expressed in all hematopoietic tissues and cell lines. Highly expressed in a variety of tumors characterized by increased mitotic activity with highest expression in lung cancer

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15468 | HPA portal (live; not the pinned values)

STK31 | Unsafe / Protein

ENSG00000196335

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. stomach 1 / glandular cells: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 39/133 (29.3%); Pancreatic Adenocarcinoma (validation): 21/80 (26.2%); Lung Adenocarcinoma (validation): 24/105 (22.9%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BXU1 | HPA portal (live; not the pinned values)

STPG3 | Unsafe / RNA

ENSG00000197768

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. duodenum: 3.8 nTPM (limit 2); small intestine: 2.8 nTPM (limit 2); duodenum: 3.8 nTPM (baseline 2); small intestine: 2.8 nTPM (baseline 2)

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 14/100 (14.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 16/521 (3.1%); Rectum Adenocarcinoma (validation): 4/207 (1.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N7X2 | HPA portal (live; not the pinned values)

STT3B | Unsafe / Protein

ENSG00000163527

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. pancreatic cancer: 9/11 (81.8%); stomach cancer: 8/10 (80.0%)

Biology. Catalytic subunit of the oligosaccharyl transferase (OST) complex that catalyzes the initial transfer of a defined glycan (Glc(3)Man(9)GlcNAc(2) in eukaryotes) from the lipid carrier dolichol-pyrophosphate to an asparagine residue within an Asn-X-Ser/Thr consensus motif in nascent polypeptide chains, the first step in protein N-glycosylation (PubMed:19167329, PubMed:31296534, PubMed:31831667, PubMed:39509507). N-glycosylation occurs ... [full text in table]

Curated tissue note. Expressed in heart, brain, placenta, lung, liver, muscle, kidney and pancreas. Expressed in skin fibroblasts (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TCJ2 | HPA portal (live; not the pinned values)

STXBP4 | Unsafe / Protein

ENSG00000166263

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 424/486 (87.2%); Pancreatic Adenocarcinoma (validation): 68/80 (85.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); melanoma: 12/12 (100.0%)

Biology. Plays a role in the translocation of transport vesicles from the cytoplasm to the plasma membrane. Inhibits the translocation of SLC2A4 from intracellular vesicles to the plasma membrane by STX4A binding and preventing the interaction between STX4A and VAMP2. Stimulation with insulin disrupts the interaction with STX4A, leading to increased levels of SLC2A4 at the plasma membrane. May also play a role in the regulation of insulin ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZWJ1 | HPA portal (live; not the pinned values)

STXBP5 | Unsafe / Protein

ENSG00000164506

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / ciliated cells (cell body): Medium (Enhanced); bronchus / ciliated cells (ciliary rootlets): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); caudate / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. pancreatic cancer: 4/12 (33.3%); ovarian cancer: 3/10 (30.0%)

Biology. Plays a regulatory role in calcium-dependent exocytosis and neurotransmitter release. Inhibits membrane fusion between transport vesicles and the plasma membrane. May modulate the assembly of trans-SNARE complexes between transport vesicles and the plasma membrane. Inhibits translocation of GLUT4 from intracellular vesicles to the plasma membrane. Competes with STXBP1 for STX1 binding (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T5C0 | HPA portal (live; not the pinned values)

SUN3 | Unsafe / Protein

ENSG00000164744

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); skin 1 / keratinocytes: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 10/133 (7.5%); Uterine Corpus Endometrial Carcinoma (TCGA): 8/176 (4.5%); Kidney Chromophobe (TCGA): 2/64 (3.1%)

Cancer IHC. breast cancer: 0/9 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. As a probable component of the LINC (LInker of Nucleoskeleton and Cytoskeleton) complex, involved in the connection between the nuclear lamina and the cytoskeleton. The nucleocytoplasmic interactions established by the LINC complex play an important role in the transmission of mechanical forces across the nuclear envelope and in nuclear movement and positioning. May be involved in nuclear remodeling during sperm head formation in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TAQ9 | HPA portal (live; not the pinned values)

SUV39H2 | Unsafe / RNA

ENSG00000152455

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 3.3 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 471/486 (96.9%); Rectum Adenocarcinoma (validation): 200/207 (96.6%)

Cancer IHC. breast cancer: 2/9 (22.2%); stomach cancer: 1/11 (9.1%)

Biology. Histone methyltransferase that specifically mediates trimethylation of 'Lys-9' of histone H3 (H3K9me3) using monomethylated H3 'Lys-9' (H3K9me1) as substrate (PubMed:10949293). H3 'Lys-9' trimethylation represents a specific tag for epigenetic transcriptional repression by recruiting HP1 (CBX1, CBX3 and/or CBX5) proteins to methylated histones (PubMed:10949293). Mainly functions in heterochromatin regions, thereby playing a central ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H5I1 | HPA portal (live; not the pinned values)

SVOPL | Unsafe / RNA

ENSG00000157703

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 8.8 nTPM (limit 2); kidney: 2.7 nTPM (limit 2); retina: 3.1 nTPM (limit 2); choroid plexus: 8.8 nTPM (baseline 2); kidney: 2.7 nTPM (baseline 2); retina: 3.1 nTPM (baseline 2)

Cancer RNA. Kidney Chromophobe (TCGA): 43/64 (67.2%); Ovary Serous Cystadenocarcinoma (validation): 28/81 (34.6%); Ovary Serous Cystadenocarcinoma (TCGA): 78/349 (22.3%)

Cancer IHC. glioma: 11/12 (91.7%); liver cancer: 11/12 (91.7%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N434 | HPA portal (live; not the pinned values)

SYCE1 | Unsafe / RNA

ENSG00000171772

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 9.8 nTPM (limit 2); basal ganglia: 11.2 nTPM (limit 2); cerebellum: 17.5 nTPM (limit 12); cerebral cortex: 16.3 nTPM (limit 12); hippocampal formation: 8.9 nTPM (limit 2); hypothalamus: 7.3 nTPM (limit 2); midbrain: 3 nTPM (limit 2); retina: 2.2 nTPM (limit 2); spinal cord: 4 nTPM (limit ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 53/133 (39.8%); Liver Hepatocellular Carcinoma (TCGA): 45/362 (12.4%); Prostate Adenocarcinoma (TCGA): 59/480 (12.3%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Major component of the transverse central element of synaptonemal complexes (SCS), formed between homologous chromosomes during meiotic prophase. Requires SYCP1 in order to be incorporated into the central element. May have a role in the synaptonemal complex assembly, stabilization and recombination

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N0S2 | HPA portal (live; not the pinned values)

SYCE1L | Unsafe / RNA

ENSG00000205078

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.7 nTPM (limit 2); cerebellum: 2.4 nTPM (limit 2); hippocampal formation: 2.5 nTPM (limit 2); hypothalamus: 3.1 nTPM (limit 2); kidney: 3.1 nTPM (limit 2); pituitary gland: 2.1 nTPM (limit 2); retina: 2.7 nTPM (limit 2); small intestine: 2.1 nTPM (limit 2); spinal cord: 2.2 nTPM (limit ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 46/50 (92.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 188/282 (66.7%); Prostate Adenocarcinoma (TCGA): 289/480 (60.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in meiosis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A8MT33 | HPA portal (live; not the pinned values)

SYCE3 | Unsafe / RNA

ENSG00000217442

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.2 nTPM (limit 2); basal ganglia: 2.4 nTPM (limit 2); kidney: 2.7 nTPM (limit 2); midbrain: 2.5 nTPM (limit 2); amygdala: 2.2 nTPM (baseline 2); basal ganglia: 2.4 nTPM (baseline 2); kidney: 2.7 nTPM (baseline 2); midbrain: 2.5 nTPM (baseline 2)

Cancer RNA. Kidney Renal Papillary Cell Carcinoma (TCGA): 104/282 (36.9%); Breast Invasive Carcinoma (TCGA): 292/1022 (28.6%); Thyroid Carcinoma (TCGA): 137/495 (27.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Major component of the transverse central element of synaptonemal complexes (SCS), formed between homologous chromosomes during meiotic prophase (PubMed:36635604). Required for the assembly of the central element of the synaptonemal complex during meiosis, via remodeling of SYCP1 lattice structures and promoting recruitment of SYCE2-TEX12 and SYCE1-SIX60S1 complexes (PubMed:36635604). Required for chromosome loading of the central ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A1L190 | HPA portal (live; not the pinned values)

SYCN | Unsafe / Protein

ENSG00000179751

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. pancreas / exocrine glandular cells: High (Enhanced)

Cancer RNA. Pancreatic Adenocarcinoma (TCGA): 89/176 (50.6%); Pancreatic Adenocarcinoma (validation): 11/80 (13.8%); Testicular Germ Cell Tumor (TCGA): 6/133 (4.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Functions in exocytosis in pancreatic acinar cells regulating the fusion of zymogen granules with each other. May have a pore-forming activity on membranes and regulate exocytosis in other exocrine tissues (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q0VAF6 | HPA portal (live; not the pinned values)

SYCP2 | Unsafe / RNA

ENSG00000196074

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.2 nTPM (limit 2); bone marrow: 2.3 nTPM (limit 2); cerebellum: 2.7 nTPM (limit 2); cerebral cortex: 2.8 nTPM (limit 2); heart muscle: 2.4 nTPM (limit 2); kidney: 4.2 nTPM (limit 2); lung: 2.1 nTPM (limit 2); pancreas: 6.4 nTPM (limit 2); small intestine: 2.3 nTPM (limit 2); basal ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 46/50 (92.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 231/283 (81.6%); Breast Invasive Carcinoma (TCGA): 809/1022 (79.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Major component of the axial/lateral elements of synaptonemal complexes (SCS) during meiotic prophase. Plays a role in the assembly of synaptonemal complexes. Required for normal meiotic chromosome synapsis during oocyte and spermatocyte development and for normal male and female fertility. Required for insertion of SYCP3 into synaptonemal complexes. May be involved in the organization of chromatin by temporarily binding to DNA ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BX26 | HPA portal (live; not the pinned values)

SYCP2L | Unsafe / RNA

ENSG00000153157

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. parathyroid gland: 66.4 nTPM (limit 6); parathyroid gland: 66.4 nTPM (baseline 6)

Cancer RNA. Glioblastoma Multiforme (validation): 10/58 (17.2%); Testicular Germ Cell Tumor (TCGA): 19/133 (14.3%); Breast Invasive Carcinoma (validation): 7/50 (14.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Oocyte-specific protein that localizes to centromeres at the dictyate stage and regulates the survival of primordial oocytes

Curated tissue note. Expressed in the ovary (at protein level) (PubMed:17374641). In the ovaries, expressed in oocytes, including germinal vesicle oocytes, metaphase I and metaphase II oocytes (PubMed:32303603). Also expressed in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T4T6 | HPA portal (live; not the pinned values)

SYN3 | Unsafe / Protein

ENSG00000185666

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebellum / Purkinje cells: Low (Uncertain); cerebral cortex / neuropil: Medium (Uncertain); thyroid gland / glandular cells: Low (Uncertain)

Cancer RNA. Rectum Adenocarcinoma (validation): 190/207 (91.8%); Colon Adenocarcinoma (validation): 427/486 (87.9%); Testicular Germ Cell Tumor (TCGA): 89/133 (66.9%)

Cancer IHC. endometrial cancer: 4/11 (36.4%); lung cancer: 4/12 (33.3%)

Biology. May be involved in the regulation of neurotransmitter release and synaptogenesis

Curated tissue note. Neuron specific. Detected predominantly in brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14994 | HPA portal (live; not the pinned values)

SYNGR4 | Unsafe / Protein

ENSG00000105467

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / glandular cells: Low (Enhanced); duodenum / glandular cells: Low (Enhanced); liver / hepatocytes: Low (Enhanced); rectum / glandular cells: Low (Enhanced); small intestine / glandular cells: Low (Enhanced)

Cancer RNA. Pancreatic Adenocarcinoma (TCGA): 26/176 (14.8%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 15/283 (5.3%); Lung Adenocarcinoma (TCGA): 23/497 (4.6%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95473 | HPA portal (live; not the pinned values)

SYNJ1 | Unsafe / Protein

ENSG00000159082

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Low (Enhanced); cerebellum / GLUC cells - cytoplasm/membrane: High ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 205/207 (99.0%); Colon Adenocarcinoma (validation): 476/486 (97.9%)

Cancer IHC. liver cancer: 3/11 (27.3%); glioma: 2/12 (16.7%)

Biology. Phosphatase that hydrolyzes phosphate groups from the inositol ring of phosphoinositides and inositol phosphates, in a domain-specific manner (PubMed:10224048, PubMed:18093523, PubMed:23804563, PubMed:27435091, PubMed:33349335, PubMed:40969890). The 5-PPase domain catalyzes removal of the 5-phosphate from substrates such as phosphatidylinositol-4,5-bisphosphate (PtdIns(4,5)P2), phosphatidylinositol-3,4,5-trisphosphate ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43426 | HPA portal (live; not the pinned values)

SYT10 | Unsafe / High RNA

ENSG00000110975

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 106 nTPM; vascular/schwann cells; hpa_single_cell_tissue; vital; 0.1213 TPM; blood/TREG_NAIVE; dice_treg_naive; vital

Cancer RNA. Kidney Chromophobe (TCGA): 15/64 (23.4%); Breast Invasive Carcinoma (validation): 3/50 (6.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 11/282 (3.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Ca(2+) sensor specifically required for the Ca(2+)-dependent exocytosis of secretory vesicles containing IGF1 in neurons of the olfactory bulb. Exocytosis of IGF1 is required for sensory perception of smell. Not involved in Ca(2+)-dependent synaptic vesicle exocytosis (By similarity). Acts through Ca(2+) and phospholipid binding to the C2 domain: Ca(2+) induces binding of the C2-domains to phospholipid membranes and to assembled ... [full text in table]

Curated tissue note. Expressed only in pancreas, lung and kidney

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6XYQ8 | HPA portal (live; not the pinned values)

SYT14 | Unsafe / RNA

ENSG00000143469

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 3.8 nTPM (limit 2); retina: 3.8 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 45/58 (77.6%); Skin Cuteneous Melanoma (TCGA): 3/99 (3.0%); Lung Squamous Cell Carcinoma (validation): 2/68 (2.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in the trafficking and exocytosis of secretory vesicles in non-neuronal tissues. Is Ca(2+)-independent

Curated tissue note. Highly expressed in fetal and adult brain tissue

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NB59 | HPA portal (live; not the pinned values)

TACC3 | Unsafe / Protein

ENSG00000013810

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); colon / glandular cells: Medium (Enhanced); duodenum / glandular cells: Medium (Enhanced); esophagus / squamous epithelial cells: Low (Enhanced); kidney / distal tubules: Medium (Enhanced); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%); Rectum Adenocarcinoma (TCGA): 88/88 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); cervical cancer: 11/12 (91.7%)

Biology. Plays a role in the microtubule-dependent coupling of the nucleus and the centrosome. Involved in the processes that regulate centrosome-mediated interkinetic nuclear migration (INM) of neural progenitors (By similarity). Acts as a component of the TACC3/ch-TOG/clathrin complex proposed to contribute to stabilization of kinetochore fibers of the mitotic spindle by acting as inter-microtubule bridge. The TACC3/ch-TOG/clathrin complex is ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y6A5 | HPA portal (live; not the pinned values)

TADA2A | Unsafe / RNA

ENSG00000276234

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.4 nTPM (limit 2); amygdala: 5.8 nTPM (limit 2); basal ganglia: 6.2 nTPM (limit 2); bone marrow: 2.5 nTPM (limit 2); cerebellum: 10.2 nTPM (limit 2); cerebral cortex: 7.6 nTPM (limit 2); choroid plexus: 2.5 nTPM (limit 2); heart muscle: 4.1 nTPM (limit 2); hippocampal formation: 5.5 ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 45/50 (90.0%); Rectum Adenocarcinoma (validation): 182/207 (87.9%); Colon Adenocarcinoma (validation): 404/486 (83.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

HPA portal (live; not the pinned values)

TAF1A | Unsafe / Protein

ENSG00000143498

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular layer: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 50/50 (100.0%); Pancreatic Adenocarcinoma (validation): 79/80 (98.8%)

Cancer IHC. testis cancer: 10/10 (100.0%); colorectal cancer: 10/11 (90.9%)

Biology. Component of the transcription factor SL1/TIF-IB complex, which is involved in the assembly of the PIC (pre-initiation complex) during RNA polymerase I-dependent transcription. The rate of PIC formation probably is primarily dependent on the rate of association of SL1/TIF-IB with the rDNA promoter. SL1/TIF-IB is involved in stabilization of nucleolar transcription factor 1/UBTF on rDNA. Formation of SL1/TIF-IB excludes the association ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15573 | HPA portal (live; not the pinned values)

TAF4B | Unsafe / Protein

ENSG00000141384

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebellum / cells in molecular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 53/58 (91.4%); Breast Invasive Carcinoma (validation): 42/50 (84.0%); Colon Adenocarcinoma (validation): 296/486 (60.9%)

Cancer IHC. colorectal cancer: 6/12 (50.0%); ovarian cancer: 3/12 (25.0%)

Biology. Cell type-specific subunit of the general transcription factor TFIID that may function as a gene-selective coactivator in certain cells. TFIID is a multimeric protein complex that plays a central role in mediating promoter responses to various activators and repressors. TAF4B is a transcriptional coactivator of the p65/RELA NF-kappa-B subunit. Involved in the activation of a subset of antiapoptotic genes including TNFAIP3. May be ... [full text in table]

Curated tissue note. Preferentially expressed in ovarian granulosa cells (at protein level). Highly expressed in B-cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q92750 | HPA portal (live; not the pinned values)

TAF7L | Unsafe / RNA

ENSG00000102387

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3.9 nTPM (limit 2)

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 79/283 (27.9%); Kidney Chromophobe (TCGA): 6/64 (9.4%); Skin Cuteneous Melanoma (TCGA): 7/99 (7.1%)

Cancer IHC. melanoma: 8/11 (72.7%); colorectal cancer: 7/11 (63.6%)

Biology. Probably functions as a spermatogenesis-specific component of the DNA-binding general transcription factor complex TFIID, a multimeric protein complex that plays a central role in mediating promoter responses to various activators and repressors. May play a role in spermatogenesis (By similarity)

Curated tissue note. Testis-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5H9L4 | HPA portal (live; not the pinned values)

TAFA3 | Unsafe / Protein

ENSG00000184599

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 15/99 (15.2%); Glioblastoma Multiforme (TCGA): 17/141 (12.1%); Glioblastoma Multiforme (validation): 6/58 (10.3%)

Cancer IHC. melanoma: 5/9 (55.6%); renal cancer: 4/12 (33.3%)

Biology. Plays a role in the regulation of microglia polarization

Curated tissue note. Brain-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z5A8 | HPA portal (live; not the pinned values)

TATDN2 | Unsafe / Protein

ENSG00000157014

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. ovarian cancer: 11/11 (100.0%); cervical cancer: 10/11 (90.9%)

Biology. Mg(2+)-dependent 3'RNA exonuclease and endonuclease that resolves R-loops via specific degradation of R-loop RNA structure (PubMed:37953292). Shows no activity against D-loop and minimal activity against the RNA strand of an RNA-DNA hybrid duplex oligomer. Has no 3' or 5' exonuclease activity, no uracil glycosylase activity, and no 5' flap endonuclease activity on DNA substrates (PubMed:37953292). May have a role in maintaining genomic ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q93075 | HPA portal (live; not the pinned values)

TBATA | Unsafe / RNA

ENSG00000166220

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. thymus: 125.3 nTPM (limit 10); thymus: 125.3 nTPM (baseline 10)

Cancer RNA. Kidney Chromophobe (TCGA): 10/64 (15.6%); Ovary Serous Cystadenocarcinoma (validation): 1/81 (1.2%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May play a role in spermatid differentiation. Modulates thymic stromal cell proliferation and thymus function

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96M53 | HPA portal (live; not the pinned values)

TBC1D26 | Unsafe / RNA

ENSG00000214946

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 4.2 nTPM (limit 2); hippocampal formation: 3 nTPM (limit 2); basal ganglia: 4.2 nTPM (baseline 2); hippocampal formation: 3 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Lung Squamous Cell Carcinoma (TCGA): 1/489 (0.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May act as a GTPase-activating protein for Rab family protein(s)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86UD7 | HPA portal (live; not the pinned values)

TBC1D3D | Unsafe / Protein

ENSG00000274419

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. pancreas / exocrine glandular cells: Low (Approved); urinary bladder / urothelial cells: Low (Approved)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 26/176 (14.8%); Kidney Renal Papillary Cell Carcinoma (TCGA): 38/282 (13.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 30/283 (10.6%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Acts as a GTPase activating protein for RAB5. Does not act on RAB4 or RAB11 (By similarity)

Curated tissue note. Expressed in pancreas, thymus and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0A087WVF3 | HPA portal (live; not the pinned values)

TBC1D3G | Unsafe / Protein

ENSG00000260287

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / cells in tubules: Low (Supported)

Cancer RNA. Breast Invasive Carcinoma (validation): 17/50 (34.0%); Bladder Urothelial Carcinoma (TCGA): 2/169 (1.2%); Glioblastoma Multiforme (TCGA): 1/141 (0.7%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Acts as a GTPase activating protein for RAB5. Does not act on RAB4 or RAB11 (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6DHY5 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

TBC1D7 | Unsafe / Protein

ENSG00000145979

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); caudate / glial cells: Low (Approved); cerebral cortex / glial cells: Medium (Approved); duodenum / glandular cells: Low (Approved); hippocampus / glial cells: Low (Approved); kidney / cells in tubules: High (Approved); liver / hepatocytes: High ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%)

Cancer IHC. prostate cancer: 6/12 (50.0%); liver cancer: 5/11 (45.5%)

Biology. Non-catalytic component of the TSC-TBC complex, a multiprotein complex that acts as a negative regulator of the canonical mTORC1 complex, an evolutionarily conserved central nutrient sensor that stimulates anabolic reactions and macromolecule biosynthesis to promote cellular biomass generation and growth (PubMed:22795129, PubMed:24529379). The TSC-TBC complex acts as a GTPase-activating protein (GAP) for the small GTPase RHEB, a direct ... [full text in table]

Curated tissue note. Highly expressed in heart, and slightly in kidney, liver and placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P0N9 | HPA portal (live; not the pinned values)

TBCE | Unsafe / Protein

ENSG00000284770

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 95/105 (90.5%); Lung Squamous Cell Carcinoma (validation): 60/68 (88.2%); Breast Invasive Carcinoma (TCGA): 792/1022 (77.5%)

Cancer IHC. colorectal cancer: 10/12 (83.3%); liver cancer: 7/12 (58.3%)

Biology. Tubulin-folding protein; involved in the second step of the tubulin folding pathway and in the regulation of tubulin heterodimer dissociation. Required for correct organization of microtubule cytoskeleton and mitotic splindle, and maintenance of the neuronal microtubule network

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15813 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

TBCEL | Unsafe / Protein

ENSG00000154114

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / ciliated cells (cell body): Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / Purkinje cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Adenocarcinoma (validation): 103/105 (98.1%); Lung Squamous Cell Carcinoma (validation): 65/68 (95.6%)

Cancer IHC. breast cancer: 12/12 (100.0%); ovarian cancer: 11/11 (100.0%)

Biology. Acts as a regulator of tubulin stability

Curated tissue note. Abundantly expressed in testis, but is also present in several tissues at a much lower level

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5QJ74 | HPA portal (live; not the pinned values)

TBL2 | Unsafe / RNA

ENSG00000106638

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 7.5 nTPM (limit 2); heart muscle: 3.2 nTPM (limit 2); kidney: 3.7 nTPM (limit 2); liver: 4.5 nTPM (limit 2); lung: 4 nTPM (limit 2); pancreas: 15.5 nTPM (limit 2); pituitary gland: 2.9 nTPM (limit 2); small intestine: 3.9 nTPM (limit 2); adrenal gland: 7.5 nTPM (baseline 2); heart ... [full text in table]

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Ovary Serous Cystadenocarcinoma (validation): 81/81 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y4P3 | HPA portal (live; not the pinned values)

TCEAL1 | Unsafe / Protein

ENSG00000172465

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: High (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / goblet cells: High (Approved); caudate / neuronal cells: Low (Approved); cerebral ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. cervical cancer: 10/11 (90.9%); melanoma: 10/11 (90.9%)

Biology. May be involved in transcriptional regulation. Modulates various viral and cellular promoters in a promoter context-dependent manner. For example, transcription from the FOS promoter is increased, while Rous sarcoma virus (RSV) long terminal repeat (LTR) promoter activity is repressed. Does not bind DNA directly

Curated tissue note. Expressed in all tissues examined. Highly expressed in heart, ovary, prostate and skeletal muscle. Moderately expressed in brain, placenta, testis and small intestine. Weakly expressed in lung, liver and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15170 | HPA portal (live; not the pinned values)

TCL1B | Unsafe / High RNA

ENSG00000213231

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 44.69 nTPM; blood/naive B-cell; hpa_immune_cell_samples; vital; 34.46 TPM; blood/naive B-cell; hpa_immune_cell_samples; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 100/133 (75.2%); Breast Invasive Carcinoma (validation): 4/50 (8.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 9/176 (5.1%)

Cancer IHC. prostate cancer: 6/11 (54.5%); glioma: 5/10 (50.0%)

Biology. Enhances the phosphorylation and activation of AKT1 and AKT2

Curated tissue note. Expressed in a variety of tissues including placenta and testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95988 | HPA portal (live; not the pinned values)

TCP10L | Unsafe / Protein

ENSG00000242220

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. liver / hepatocytes: Medium (Approved)

Cancer RNA. Liver Hepatocellular Carcinoma (TCGA): 154/362 (42.5%); Glioblastoma Multiforme (validation): 19/58 (32.8%); Liver Hepatocellular Carcinoma (validation): 39/231 (16.9%)

Cancer IHC. liver cancer: 4/12 (33.3%); testis cancer: 1/9 (11.1%)

Biology. May be involved in transcriptional regulation. Has in vitro transcription inhibition activity. Acts as a tumor suppressor in hepatocellular carcinoma (HCC) cells

Curated tissue note. Expressed in liver and testis. Expressed in the seminiferous tubules (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TDR4 | HPA portal (live; not the pinned values)

TCP11L1 | Unsafe / Protein

ENSG00000176148

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 483/486 (99.4%)

Cancer IHC. cervical cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NUJ3 | HPA portal (live; not the pinned values)

TCTE1 | Unsafe / RNA

ENSG00000146221

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.1 nTPM (limit 2); basal ganglia: 2.6 nTPM (limit 2); cerebellum: 7.6 nTPM (limit 2); cerebral cortex: 2.7 nTPM (limit 2); hippocampal formation: 4.3 nTPM (limit 2); hypothalamus: 2.8 nTPM (limit 2); amygdala: 3.1 nTPM (baseline 2); basal ganglia: 2.6 nTPM (baseline 2); cerebellum: 7.6 ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 26/176 (14.8%); Testicular Germ Cell Tumor (TCGA): 6/133 (4.5%); Lung Adenocarcinoma (TCGA): 11/497 (2.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of the nexin-dynein regulatory complex (N-DRC) a key regulator of ciliary/flagellar motility which maintains the alignment and integrity of the distal axoneme and regulates microtubule sliding in motile axonemes. May play a role in the assembly of N-DRC. May be required for sperm motility

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JU00 | HPA portal (live; not the pinned values)

TDRD10 | Unsafe / RNA

ENSG00000163239

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 10.7 nTPM (limit 10); adrenal gland: 4.3 nTPM (limit 2); heart muscle: 7.4 nTPM (limit 2); kidney: 3.4 nTPM (limit 2); liver: 4.4 nTPM (limit 2); lung: 7.8 nTPM (limit 2); pituitary gland: 2.5 nTPM (limit 2); small intestine: 3.4 nTPM (limit 2); thyroid gland: 10.7 nTPM (limit 10); ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 55/133 (41.4%); Liver Hepatocellular Carcinoma (validation): 82/231 (35.5%); Breast Invasive Carcinoma (validation): 17/50 (34.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5VZ19 | HPA portal (live; not the pinned values)

TDRD5 | Unsafe / Protein

ENSG00000162782

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / collecting ducts: Medium (Enhanced); kidney / distal tubules: Medium (Enhanced); kidney / proximal tubules (cell body): Medium (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 45/50 (90.0%); Breast Invasive Carcinoma (TCGA): 487/1022 (47.7%); Lung Squamous Cell Carcinoma (TCGA): 128/489 (26.2%)

Cancer IHC. liver cancer: 2/9 (22.2%); breast cancer: 0/9 (0.0%)

Biology. Required during spermiogenesis to participate in the repression transposable elements and prevent their mobilization, which is essential for the germline integrity. Probably acts via the piRNA metabolic process, which mediates the repression of transposable elements during meiosis by forming complexes composed of piRNAs and Piwi proteins and govern the methylation and subsequent repression of transposons. Required for chromatoid body ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NAT2 | HPA portal (live; not the pinned values)

TDRD6 | Unsafe / RNA

ENSG00000180113

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3 nTPM (limit 2); pituitary gland: 2.9 nTPM (limit 2); retina: 8.6 nTPM (limit 2)

Cancer RNA. Prostate Adenocarcinoma (TCGA): 254/480 (52.9%); Lung Adenocarcinoma (validation): 23/105 (21.9%); Lung Squamous Cell Carcinoma (validation): 10/68 (14.7%)

Cancer IHC. breast cancer: 2/12 (16.7%); liver cancer: 1/10 (10.0%)

Biology. Tudor domain-containing protein involved in germ cell development, more specifically the formation of chromatoid body (during spermiogenesis), Balbiani body (during oogenesis), germ plasm (upon fertilization), and for proper miRNA expression and spliceosome maturation (By similarity). Essential for RNA-dependent helicase UPF1 localization to chromatoid body, for UPF1-UPF2 and UPF1-DDX4 interactions which are required for mRNA ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O60522 | HPA portal (live; not the pinned values)

TDRKH | Unsafe / Protein

ENSG00000182134

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / Purkinje cells: Low ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 105/105 (100.0%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 48/50 (96.0%)

Cancer IHC. renal cancer: 10/12 (83.3%); ovarian cancer: 8/12 (66.7%)

Biology. Participates in the primary piRNA biogenesis pathway and is required during spermatogenesis to repress transposable elements and prevent their mobilization, which is essential for the germline integrity. The piRNA metabolic process mediates the repression of transposable elements during meiosis by forming complexes composed of piRNAs and Piwi proteins and govern the methylation and subsequent repression of transposons. Required for the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y2W6 | HPA portal (live; not the pinned values)

TECTA | Unsafe / Protein

ENSG00000109927

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); colon / endothelial cells: Low (Approved); colon / peripheral nerve/ganglion: Low (Approved); duodenum / glandular cells: Low (Approved); esophagus / squamous epithelial cells: Low (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 14/58 (24.1%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Glioblastoma Multiforme (TCGA): 1/141 (0.7%)

Cancer IHC. testis cancer: 2/11 (18.2%); cervical cancer: 2/12 (16.7%)

Biology. One of the major non-collagenous components of the tectorial membrane (By similarity). The tectorial membrane is an extracellular matrix of the inner ear that covers the neuroepithelium of the cochlea and contacts the stereocilia bundles of specialized sensory hair cells. Sound induces movement of these hair cells relative to the tectorial membrane, deflects the stereocilia and leads to fluctuations in hair-cell membrane potential, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75443 | HPA portal (live; not the pinned values)

TEDC2 | Unsafe / Protein

ENSG00000162062

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. duodenum / glandular cells: Low (Approved); hippocampus / glial cells: Low (Approved); kidney / cells in tubules: Low (Approved); lung / macrophages: Low (Approved); rectum / glandular cells: Low (Approved); skeletal muscle / myocytes: Low (Approved); spleen / cells in red pulp: Low (Approved); ... [full text in table]

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 258/283 (91.2%); Testicular Germ Cell Tumor (TCGA): 114/133 (85.7%); Skin Cuteneous Melanoma (TCGA): 76/99 (76.8%)

Cancer IHC. lung cancer: 4/12 (33.3%); skin cancer: 2/12 (16.7%)

Biology. Acts as a positive regulator of ciliary hedgehog signaling. Required for centriole stability

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7L2K0 | HPA portal (live; not the pinned values)

TEKT3 | Unsafe / Protein

ENSG00000125409

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): Medium (Enhanced); bronchus / ciliated cells (tip of cilia): Medium (Enhanced); lung / macrophages: Low (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 6/50 (12.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 11/176 (6.2%); Testicular Germ Cell Tumor (TCGA): 7/133 (5.3%)

Cancer IHC. renal cancer: 3/12 (25.0%); colorectal cancer: 2/12 (16.7%)

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia and flagellar axoneme (PubMed:36191189). Forms filamentous polymers in the walls of ciliary and flagellar microtubules (By similarity). Required for normal sperm mobility (By similarity)

Curated tissue note. Expressed in spermatozoa (PubMed:36708031). Expressed in airway epithelial cells (PubMed:36191189)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BXF9 | HPA portal (live; not the pinned values)

TEKT4 | Unsafe / Protein

ENSG00000163060

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cilia axoneme): High (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 8/50 (16.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 27/176 (15.3%); Ovary Serous Cystadenocarcinoma (TCGA): 16/349 (4.6%)

Cancer IHC. endometrial cancer: 5/11 (45.5%); urothelial cancer: 5/11 (45.5%)

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia and flagellar axoneme (PubMed:36191189). Forms filamentous polymers in the walls of ciliary and flagellar microtubules (By similarity). Contributes to normal sperm motility (By similarity)

Curated tissue note. Strongly expressed in spermatozoa. Also detected at low levels in pancreas. Expressed in airway epithelial cells (PubMed:36191189)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WW24 | HPA portal (live; not the pinned values)

TEKTL1 | Unsafe / Protein

ENSG00000160994

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); colon / glandular cells: Low (Approved); duodenum / glandular cells: Low (Approved); esophagus / squamous epithelial cells: Medium (Approved); kidney / cells in tubules: Low (Approved); ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Ovary Serous Cystadenocarcinoma (TCGA): 1/349 (0.3%)

Cancer IHC. testis cancer: 3/11 (27.3%); lymphoma: 1/11 (9.1%)

Biology. Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in sperm flagellar axoneme, which is required for motile flagellum beating (By similarity). Forms an extensive interaction network cross-linking the lumen of axonemal doublet microtubules (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYK2 | HPA portal (live; not the pinned values)

TESMIN | Unsafe / RNA

ENSG00000132749

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 7.4 nTPM (limit 2); kidney: 3 nTPM (limit 2); pituitary gland: 2.3 nTPM (limit 2); bone marrow: 7.4 nTPM (baseline 2); kidney: 3 nTPM (baseline 2); pituitary gland: 2.3 nTPM (baseline 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (validation): 73/81 (90.1%); Breast Invasive Carcinoma (TCGA): 880/1022 (86.1%); Kidney Chromophobe (TCGA): 48/64 (75.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Essential for normal spermatogenesis and male fertility (By similarity). Required for the completion of meiosis in male germ cells (By similarity)

Curated tissue note. Expressed specifically in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y4I5 | HPA portal (live; not the pinned values)

TET1 | Unsafe / High RNA

ENSG00000138336

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 51.7 nTPM; unsited/Astrocytes; hpa_single_cell; unresolved; 8.088 TPM; blood/CD8_NAIVE; dice_cd8_naive; vital

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Testicular Germ Cell Tumor (TCGA): 108/133 (81.2%); Breast Invasive Carcinoma (validation): 7/50 (14.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Dioxygenase that plays a key role in active DNA demethylation, by catalyzing the sequential oxidation of the modified genomic base 5-methylcytosine (5mC) into 5-hydroxymethylcytosine (5hmC), 5-formylcytosine (5fC), and 5-carboxylcytosine (5caC) (PubMed:19372391, PubMed:21496894, PubMed:21778364, PubMed:35798741). In addition to its role in DNA demethylation, plays a more general role in chromatin regulation by recruiting histone ... [full text in table]

Curated tissue note. Expressed in fetal heart, lung and brain, and in adult skeletal muscle, thymus and ovary. Not detected in adult heart, lung or brain. Up-regulated in glioblastoma cells (at protein level) (PubMed:25284789) ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NFU7 | HPA portal (live; not the pinned values)

TEX101 | Unsafe / High RNA

ENSG00000131126

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 119.7 nTPM; blood/basophil; hpa_immune_cell_samples; vital; 74.9 TPM; blood/basophil; hpa_immune_monaco; vital

Cancer RNA. Lung Squamous Cell Carcinoma (TCGA): 17/489 (3.5%); Ovary Serous Cystadenocarcinoma (TCGA): 12/349 (3.4%); Stomach Adenocarcinoma (TCGA): 11/346 (3.2%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Plays a role in fertilization by controlling binding of sperm to zona pellucida and migration of spermatozoa into the oviduct (By similarity). May play a role in signal transduction and promote protein tyrosine phosphorylation (By similarity)

Curated tissue note. Detected in testis and spermatogonia. Not detected in spermatocytes. Detected in blood leukocytes

Reconsideration. Resolve basophil RNA identity and local protein evidence before reconsideration.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BY14 | HPA portal (live; not the pinned values)

TEX11 | Unsafe / RNA

ENSG00000120498

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 13.6 nTPM (limit 2); pancreas: 57 nTPM (limit 4)

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 49/100 (49.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 171/521 (32.8%); Testicular Germ Cell Tumor (TCGA): 22/133 (16.5%)

Cancer IHC. endometrial cancer: 2/11 (18.2%); melanoma: 2/11 (18.2%)

Biology. Regulator of crossing-over during meiosis. Involved in initiation and/or maintenance of chromosome synapsis and formation of crossovers

Curated tissue note. Testis-specific. Not expressed in adult ovaries

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYF3 | HPA portal (live; not the pinned values)

TEX12 | Unsafe / RNA

ENSG00000150783

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 8.4 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 4/133 (3.0%); Bladder Urothelial Carcinoma (TCGA): 2/169 (1.2%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 3/283 (1.1%)

Cancer IHC. endometrial cancer: 1/12 (8.3%); breast cancer: 0/11 (0.0%)

Biology. Component of the transverse central element of synaptonemal complexes (SCS), formed between homologous chromosomes during meiotic prophase (By similarity). Requires SYCP1 in order to be incorporated into the central element (By similarity)

Curated tissue note. Testis specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BXU0 | HPA portal (live; not the pinned values)

TEX26 | Unsafe / RNA

ENSG00000175664

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 11.6 nTPM (limit 2); hypothalamus: 3.4 nTPM (limit 2); choroid plexus: 11.6 nTPM (baseline 2); hypothalamus: 3.4 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 32/58 (55.2%); Glioblastoma Multiforme (TCGA): 37/141 (26.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 5/176 (2.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N6G2 | HPA portal (live; not the pinned values)

TEX29 | Unsafe / RNA

ENSG00000153495

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 5.9 nTPM (limit 2); basal ganglia: 3.8 nTPM (limit 2); cerebral cortex: 12 nTPM (limit 2); amygdala: 5.9 nTPM (baseline 2); basal ganglia: 3.8 nTPM (baseline 2); cerebral cortex: 12 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 11/58 (19.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 12/283 (4.2%); Bladder Urothelial Carcinoma (TCGA): 5/169 (3.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N6K0 | HPA portal (live; not the pinned values)

TEX30 | Unsafe / Protein

ENSG00000151287

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. skin 1 / cells in corneal layer: High (Enhanced); skin 2 / cells in corneal layer: Medium (Enhanced)

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. testis cancer: 1/11 (9.1%); liver cancer: 1/12 (8.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JUR7 | HPA portal (live; not the pinned values)

TEX54 | Unsafe / High RNA

ENSG00000283268

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 179.5 nTPM; blood/neutrophil; hpa_immune_cell_samples; vital; 127.8 TPM; blood/neutrophil; hpa_immune_cell_samples; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Prostate Adenocarcinoma (TCGA): 7/480 (1.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 3/282 (1.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Expressed in Testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0A1B0GVG6 | HPA portal (live; not the pinned values)

TEX55 | Unsafe / Protein

ENSG00000163424

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 2/176 (1.1%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Testis-specific

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96M34 | HPA portal (live; not the pinned values)

TEX9 | Unsafe / RNA

ENSG00000151575

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.1 nTPM (limit 2); amygdala: 2.8 nTPM (limit 2); basal ganglia: 3.7 nTPM (limit 2); choroid plexus: 6.8 nTPM (limit 2); duodenum: 5.4 nTPM (limit 2); hippocampal formation: 3.5 nTPM (limit 2); hypothalamus: 4.7 nTPM (limit 2); kidney: 6.3 nTPM (limit 2); lung: 2.2 nTPM (limit 2); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 485/486 (99.8%); Rectum Adenocarcinoma (validation): 206/207 (99.5%); Kidney Renal Clear Cell Carcinoma (validation): 96/100 (96.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); endometrial cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N6V9 | HPA portal (live; not the pinned values)

TFPI2 | Unsafe / RNA

ENSG00000105825

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 27.4 nTPM (limit 12); hypothalamus: 4.3 nTPM (limit 2); kidney: 19.8 nTPM (limit 12); liver: 20.6 nTPM (limit 12); pancreas: 74.1 nTPM (limit 12); pituitary gland: 9.4 nTPM (limit 2); retina: 50.1 nTPM (limit 2); urinary bladder: 25.6 nTPM (limit 24)

Cancer RNA. Kidney Renal Papillary Cell Carcinoma (TCGA): 263/282 (93.3%); Kidney Renal Clear Cell Carcinoma (TCGA): 446/521 (85.6%); Uterine Corpus Endometrial Carcinoma (TCGA): 144/176 (81.8%)

Cancer IHC. endometrial cancer: 2/10 (20.0%); lung cancer: 2/12 (16.7%)

Biology. May play a role in the regulation of plasmin-mediated matrix remodeling. Inhibits trypsin, plasmin, factor VIIa/tissue factor and weakly factor Xa. Has no effect on thrombin

Curated tissue note. Umbilical vein endothelial cells, liver, placenta, heart, pancreas, and maternal serum at advanced pregnancy

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P48307 | HPA portal (live; not the pinned values)

THADA | Unsafe / Protein

ENSG00000115970

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebral cortex / neuronal cells: Low (Uncertain); cerebral cortex / neuropil: Medium ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%)

Cancer IHC. liver cancer: 3/12 (25.0%); skin cancer: 3/12 (25.0%)

Biology. Together with methyltransferase FTSJ1, methylates the 2'-O-ribose of nucleotides at position 32 of the anticodon loop of substrate tRNAs

Curated tissue note. Expressed in pancreas, adrenal medulla, thyroid, adrenal cortex, testis, thymus, small intestine and stomach

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6YHU6 | HPA portal (live; not the pinned values)

THAP2 | Unsafe / Protein

ENSG00000173451

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: High (Approved); bronchus / ciliated cells (cell body): High (Approved); bronchus / goblet cells: High (Approved); caudate / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Squamous Cell Carcinoma (validation): 66/68 (97.1%); Lung Adenocarcinoma (validation): 99/105 (94.3%)

Cancer IHC. prostate cancer: 10/11 (90.9%); breast cancer: 7/12 (58.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0W7 | HPA portal (live; not the pinned values)

THAP6 | Unsafe / RNA

ENSG00000174796

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 11.1 nTPM (limit 2); amygdala: 5.8 nTPM (limit 2); basal ganglia: 6 nTPM (limit 2); bone marrow: 12.4 nTPM (limit 2); cerebellum: 11.4 nTPM (limit 2); cerebral cortex: 12.4 nTPM (limit 2); choroid plexus: 6.5 nTPM (limit 2); colon: 9.6 nTPM (limit 6); duodenum: 12.1 nTPM (limit 2); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. liver cancer: 6/11 (54.5%); pancreatic cancer: 2/11 (18.2%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TBB0 | HPA portal (live; not the pinned values)

THAP9 | Unsafe / Protein

ENSG00000168152

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Pancreatic Adenocarcinoma (validation): 27/80 (33.8%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Active transposase that specifically recognizes the bipartite 5'-TXXGGGX(A/T)-3' consensus motif and mediates transposition

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H5L6 | HPA portal (live; not the pinned values)

TICRR | Unsafe / RNA

ENSG00000140534

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 3.4 nTPM (limit 2); bone marrow: 3.4 nTPM (baseline 2)

Cancer RNA. Colon Adenocarcinoma (validation): 417/486 (85.8%); Rectum Adenocarcinoma (validation): 166/207 (80.2%); Testicular Germ Cell Tumor (TCGA): 92/133 (69.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Regulator of DNA replication and S/M and G2/M checkpoints. Regulates the triggering of DNA replication initiation via its interaction with TOPBP1 by participating in CDK2-mediated loading of CDC45L onto replication origins. Required for the transition from pre-replication complex (pre-RC) to pre-initiation complex (pre-IC). Required to prevent mitotic entry after treatment with ionizing radiation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z2Z1 | HPA portal (live; not the pinned values)

TIGD7 | Unsafe / Protein

ENSG00000140993

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); colon / endothelial cells: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 48/50 (96.0%); Prostate Adenocarcinoma (TCGA): 404/480 (84.2%)

Cancer IHC. colorectal cancer: 11/12 (91.7%); cervical cancer: 10/11 (90.9%)

Biology. Expressed in all tissues tested. Higher expression in testis and ovary

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6NT04 | HPA portal (live; not the pinned values)

TIMD4 | Unsafe / Protein

ENSG00000145850

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Approved); lymph node / germinal center cells: Medium (Approved); lymph node / non-germinal center cells: Medium (Approved); spleen / cells in red pulp: Medium (Approved)

Cancer RNA. Kidney Chromophobe (TCGA): 58/64 (90.6%); Kidney Renal Clear Cell Carcinoma (validation): 79/100 (79.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 184/282 (65.2%)

Cancer IHC. breast cancer: 4/12 (33.3%); prostate cancer: 4/12 (33.3%)

Biology. Phosphatidylserine receptor that plays different role in immune response including phagocytosis of apoptotic cells and T-cell regulation (PubMed:18082433, PubMed:32640697). Involved in efferocytosis, the process by which apoptotic cells are removed by phagocytic cells by promoting the engulfment of apoptotic cells or exogenous particles (PubMed:18082433, PubMed:32703939, PubMed:34067457). Mechanistically, acts by securing apoptotic ... [full text in table]

Curated tissue note. Expressed by macrophages and dendritic cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96H15 | HPA portal (live; not the pinned values)

TIPIN | Unsafe / Protein

ENSG00000075131

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); colon / glandular cells: Medium (Enhanced); duodenum / glandular cells: Low (Enhanced); esophagus / squamous epithelial cells: Low (Enhanced); lymph node / germinal center cells: Medium (Enhanced); oral mucosa / squamous epithelial cells: Medium ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 139/141 (98.6%)

Cancer IHC. cervical cancer: 10/12 (83.3%); testis cancer: 9/11 (81.8%)

Biology. Plays an important role in the control of DNA replication and the maintenance of replication fork stability (PubMed:17102137, PubMed:23359676, PubMed:35585232). Important for cell survival after DNA damage or replication stress (PubMed:17116885). May be specifically required for the ATR-CHEK1 pathway in the replication checkpoint induced by hydroxyurea or ultraviolet light (PubMed:17296725). Forms a complex with TIMELESS and this ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BVW5 | HPA portal (live; not the pinned values)

TIRAP | Unsafe / RNA

ENSG00000150455

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.3 nTPM (limit 2); cerebellum: 2.6 nTPM (limit 2); cerebral cortex: 2.8 nTPM (limit 2); choroid plexus: 3.7 nTPM (limit 2); duodenum: 7.7 nTPM (limit 2); heart muscle: 6.2 nTPM (limit 2); hypothalamus: 2.4 nTPM (limit 2); kidney: 4 nTPM (limit 2); liver: 6.6 nTPM (limit 2); lung: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 484/486 (99.6%); Rectum Adenocarcinoma (validation): 206/207 (99.5%); Glioblastoma Multiforme (validation): 57/58 (98.3%)

Cancer IHC. colorectal cancer: 4/10 (40.0%); renal cancer: 2/11 (18.2%)

Biology. Adapter involved in TLR2, TLR4 and RAGE signaling pathways in the innate immune response. Acts via IRAK2 and TRAF-6, leading to the activation of NF-kappa-B, MAPK1, MAPK3 and JNK, and resulting in cytokine secretion and the inflammatory response. Positively regulates the production of TNF (TNF) and interleukin-6 (IL6)

Curated tissue note. Highly expressed in liver, kidney, spleen, skeletal muscle and heart. Also detected in peripheral blood leukocytes, lung, placenta, small intestine, thymus, colon and brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P58753 | HPA portal (live; not the pinned values)

TKTL1 | Unsafe / RNA

ENSG00000007350

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. retina: 2.1 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 53/133 (39.8%); Glioblastoma Multiforme (validation): 12/58 (20.7%); Ovary Serous Cystadenocarcinoma (TCGA): 72/349 (20.6%)

Cancer IHC. lung cancer: 2/12 (16.7%); breast cancer: 0/11 (0.0%)

Biology. Catalyzes the transfer of a two-carbon ketol group from a ketose donor to an aldose acceptor, via a covalent intermediate with the cofactor thiamine pyrophosphate (By similarity). Thus, catalyzes the reversible transfer of a two-carbon ketol group from sedoheptulose-7-phosphate to glyceraldehyde-3-phosphate, producing xylulose-5-phosphate and ribose-5-phosphate (By similarity). The phosphate group of the substrate plays a role in ... [full text in table]

Curated tissue note. Widely expressed (PubMed:8838793). Expressed in endothelial cells and in peripheral neurons (at protein level) (PubMed:15991799) Not expressed in fetal neocortex Expressed in fetal neocortex

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P51854 | HPA portal (live; not the pinned values)

TLCD3B | Unsafe / RNA

ENSG00000149926

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 24.5 nTPM (limit 2); basal ganglia: 21.8 nTPM (limit 2); cerebellum: 85.2 nTPM (limit 2); cerebral cortex: 42 nTPM (limit 2); hippocampal formation: 30.7 nTPM (limit 2); hypothalamus: 18.5 nTPM (limit 2); midbrain: 15.8 nTPM (limit 2); pituitary gland: 20.9 nTPM (limit 2); retina: 65.8 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 101/141 (71.6%); Breast Invasive Carcinoma (TCGA): 117/1022 (11.4%); Testicular Germ Cell Tumor (TCGA): 15/133 (11.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Involved in ceramide synthesis

Curated tissue note. Expressed in testis (PubMed:33077892). Expressed in the retina with higher expression levels in the macular than in the peripheral region (PubMed:33077892)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q71RH2 | HPA portal (live; not the pinned values)

TLE4 | Unsafe / RNA

ENSG00000106829

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 15.1 nTPM (limit 2); amygdala: 11 nTPM (limit 2); basal ganglia: 26.3 nTPM (limit 2); bone marrow: 68.1 nTPM (limit 2); cerebellum: 22.6 nTPM (limit 2); cerebral cortex: 15.8 nTPM (limit 2); choroid plexus: 21.3 nTPM (limit 2); colon: 9.6 nTPM (limit 6); duodenum: 7 nTPM (limit 2); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 205/207 (99.0%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Colon Adenocarcinoma (validation): 474/486 (97.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcriptional corepressor that binds to a number of transcription factors. Inhibits the transcriptional activation mediated by PAX5, and by CTNNB1 and TCF family members in Wnt signaling. The effects of full-length TLE family members may be modulated by association with dominant-negative AES. Essential for the transcriptional repressor activity of SIX3 during retina and lens development and for SIX3 transcriptional auto-repression ... [full text in table]

Curated tissue note. In all tissues examined, mostly in brain, and muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q04727 | HPA portal (live; not the pinned values)

TLK1 | Unsafe / Protein

ENSG00000198586

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Approved); adrenal gland / glandular cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: High (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. Rapidly and transiently inhibited by phosphorylation following the generation of DNA double-stranded breaks during S-phase. This is cell cycle checkpoint and ATM-pathway dependent and appears to regulate processes involved in chromatin assembly. Isoform 3 phosphorylates and enhances the stability of the t-SNARE SNAP23, augmenting its assembly with syntaxin. Isoform 3 protects the cells from the ionizing radiation by facilitating the ... [full text in table]

Curated tissue note. Widely expressed. Present in fetal placenta, liver, kidney and pancreas but not heart or skeletal muscle. Also found in adult cell lines. Isoform 3 is ubiquitously expressed in all tissues examined

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UKI8 | HPA portal (live; not the pinned values)

TMED7 | Unsafe / Protein

ENSG00000134970

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Supported); adrenal gland / glandular cells: Low (Supported); bone marrow / hematopoietic cells: Low (Supported); bronchus / respiratory epithelial cells: Medium (Supported); caudate / glial cells: Medium (Supported); caudate / neuronal cells: Medium (Supported); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%)

Cancer IHC. endometrial cancer: 10/11 (90.9%); liver cancer: 9/12 (75.0%)

Biology. Potential role in vesicular protein trafficking, mainly in the early secretory pathway. Appears to play a role in the biosynthesis of secreted cargo including processing and post-translational modifications

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y3B3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

TMEM140 | Unsafe / RNA

ENSG00000146859

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 34.5 nTPM (limit 10); adrenal gland: 18.6 nTPM (limit 2); amygdala: 6.4 nTPM (limit 2); basal ganglia: 8.9 nTPM (limit 2); bone marrow: 10.8 nTPM (limit 2); cerebellum: 2.6 nTPM (limit 2); cerebral cortex: 8 nTPM (limit 2); choroid plexus: 5.5 nTPM (limit 2); colon: 16.6 nTPM (limit ... [full text in table]

Cancer RNA. Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Liver Hepatocellular Carcinoma (TCGA): 362/362 (100.0%); Liver Hepatocellular Carcinoma (validation): 231/231 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Expression significantly higher in gliomas than in normal brain tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NV12 | HPA portal (live; not the pinned values)

TMEM217 | Unsafe / RNA

ENSG00000172738

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.9 nTPM (limit 2); bone marrow: 2.7 nTPM (limit 2); pancreas: 2.6 nTPM (limit 2); adrenal gland: 2.9 nTPM (baseline 2); bone marrow: 2.7 nTPM (baseline 2); pancreas: 2.6 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 8/50 (16.0%); Glioblastoma Multiforme (validation): 7/58 (12.1%); Thyroid Carcinoma (TCGA): 46/495 (9.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transmembrane protein essential for sperm motility and male fertility. Forms a complex with SLC9C1 in the flagellum, which regulates sperm motility by maintaining the stability of soluble adenylyl cyclase, which produces cAMP to power flagellar movement

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N7C4 | HPA portal (live; not the pinned values)

TMEM225B | Unsafe / RNA

ENSG00000244219

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.8 nTPM (limit 2); basal ganglia: 2.5 nTPM (limit 2); cerebral cortex: 2.3 nTPM (limit 2); hippocampal formation: 2.6 nTPM (limit 2); hypothalamus: 2.7 nTPM (limit 2); kidney: 3.4 nTPM (limit 2); midbrain: 2.9 nTPM (limit 2); pituitary gland: 2.8 nTPM (limit 2); spinal cord: 2.3 nTPM ... [full text in table]

Cancer RNA. Kidney Chromophobe (TCGA): 32/64 (50.0%); Thyroid Carcinoma (TCGA): 187/495 (37.8%); Glioblastoma Multiforme (validation): 18/58 (31.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P0DP42 | HPA portal (live; not the pinned values)

TMEM232 | Unsafe / RNA

ENSG00000186952

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3.3 nTPM (limit 2); kidney: 3.5 nTPM (limit 2); retina: 3.5 nTPM (limit 2); choroid plexus: 3.3 nTPM (baseline 2); kidney: 3.5 nTPM (baseline 2); retina: 3.5 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 27/58 (46.6%); Breast Invasive Carcinoma (validation): 17/50 (34.0%); Pancreatic Adenocarcinoma (validation): 9/80 (11.2%)

Cancer IHC. breast cancer: 11/11 (100.0%); colorectal cancer: 11/12 (91.7%)

Biology. Plays a critical role for male fertility and sperm motility by regulating sperm cytoplasm removal and maintaining axoneme integrity

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt C9JQI7 | HPA portal (live; not the pinned values)

TMEM241 | Unsafe / RNA

ENSG00000134490

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.6 nTPM (limit 2); smooth muscle: 2.8 nTPM (limit 2); cerebral cortex: 2.6 nTPM (baseline 2); smooth muscle: 2.8 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Pancreatic Adenocarcinoma (validation): 70/80 (87.5%); Lung Adenocarcinoma (validation): 27/105 (25.7%)

Cancer IHC. liver cancer: 2/11 (18.2%); breast cancer: 1/11 (9.1%)

Biology. Golgi-localized UDP-N-acetylglucosamine (UDP-GlcNAc) transporter that transports UDP-N-acetylglucosamine into Golgi lumen. Contributes to lysosomal targeting of NPC2, a key protein required for lysosomal cholesterol exiting, and that utilizes the mannose-6-phosphate (M6P) modification pathway for its lysosomal targeting

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q24JQ0 | HPA portal (live; not the pinned values)

TMEM31 | Unsafe / RNA

ENSG00000179363

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. hypothalamus: 2.2 nTPM (limit 2); spinal cord: 2.2 nTPM (limit 2); hypothalamus: 2.2 nTPM (baseline 2); spinal cord: 2.2 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 25/133 (18.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 6/176 (3.4%); Kidney Chromophobe (TCGA): 2/64 (3.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JXX7 | HPA portal (live; not the pinned values)

TMEM95 | Unsafe / High RNA

ENSG00000182896

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 35.9 nTPM; esophagus/schwann cells; hpa_single_cell_tissue; protected; 0.7017 TPM; blood/CD8_STIM; dice_cd8_stim; vital

Cancer RNA. Rectum Adenocarcinoma (validation): 1/207 (0.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 1/282 (0.4%); Bladder Urothelial Carcinoma (TCGA): 0/169 (0.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Sperm protein required for fusion of sperm with the egg membrane during fertilization

Curated tissue note. Spermatozoa (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3KNT9 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

TMPPE | Unsafe / Protein

ENSG00000188167

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%); Rectum Adenocarcinoma (validation): 206/207 (99.5%)

Cancer IHC. breast cancer: 12/12 (100.0%); endometrial cancer: 12/12 (100.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZT21 | HPA portal (live; not the pinned values)

TMPRSS11F | Unsafe / RNA

ENSG00000198092

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. esophagus: 14.7 nTPM (limit 6); esophagus: 14.7 nTPM (baseline 6)

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 93/492 (18.9%); Lung Squamous Cell Carcinoma (validation): 12/68 (17.6%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 38/283 (13.4%)

Cancer IHC. prostate cancer: 11/12 (91.7%); breast cancer: 9/12 (75.0%)

Biology. Probable serine protease

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZWK6 | HPA portal (live; not the pinned values)

TMPRSS9 | Unsafe / RNA

ENSG00000178297

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. liver: 4 nTPM (limit 2); liver: 4 nTPM (baseline 2)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 39/231 (16.9%); Liver Hepatocellular Carcinoma (TCGA): 36/362 (9.9%); Skin Cuteneous Melanoma (TCGA): 8/99 (8.1%)

Cancer IHC. melanoma: 10/12 (83.3%); endometrial cancer: 2/11 (18.2%)

Biology. Serase-1 and serase-2 are serine proteases that hydrolyze the peptides N-t-Boc-Gln-Ala-Arg-AMC and N-t-Boc-Gln-Gly-Arg-AMC. In contrast, N-t-Boc-Ala-Phe-Lys-AMC and N-t-Boc-Ala-Pro-Ala-AMC are not significantly hydrolyzed

Curated tissue note. Expressed in fetal human tissues, such as kidney, liver, lung and brain, and in a variety of tumor cell lines. Weakly expressed in adult tissues including skeletal muscle, liver, placenta and heart

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z410 | HPA portal (live; not the pinned values)

TNFSF11 | Unsafe / Protein

ENSG00000120659

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); lymph node / germinal center cells: Medium (Enhanced); lymph node / non-germinal center cells: Medium (Enhanced); spleen / cells in red pulp: Medium (Enhanced)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 45/80 (56.2%); Rectum Adenocarcinoma (validation): 110/207 (53.1%); Colon Adenocarcinoma (validation): 206/486 (42.4%)

Cancer IHC. skin cancer: 1/10 (10.0%); endometrial cancer: 1/12 (8.3%)

Biology. Cytokine that binds to TNFRSF11B/OPG and to TNFRSF11A/RANK. Osteoclast differentiation and activation factor. Augments the ability of dendritic cells to stimulate naive T-cell proliferation. May be an important regulator of interactions between T-cells and dendritic cells and may play a role in the regulation of the T-cell-dependent immune response. May also play an important role in enhanced bone-resorption in humoral hypercalcemia of ... [full text in table]

Curated tissue note. Highest in the peripheral lymph nodes, weak in spleen, peripheral blood Leukocytes, bone marrow, heart, placenta, skeletal muscle, stomach and thyroid

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14788 | HPA portal (live; not the pinned values)

TNFSF4 | Unsafe / Protein

ENSG00000117586

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / ciliated cells (cell body): Medium (Approved); bronchus / ciliated cells (ciliary rootlets): Medium (Approved); bronchus / ciliated cells (tip of cilia): Medium (Approved); caudate / ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 37/50 (74.0%); Glioblastoma Multiforme (validation): 33/58 (56.9%); Pancreatic Adenocarcinoma (validation): 44/80 (55.0%)

Cancer IHC. prostate cancer: 8/11 (72.7%); liver cancer: 7/12 (58.3%)

Biology. Cytokine that binds to TNFRSF4. Co-stimulates T-cell proliferation and cytokine production

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P23510 | HPA portal (live; not the pinned values)

TNP1 | Unsafe / High RNA

ENSG00000118245

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 250.5 nTPM; testis/macrophages; hpa_single_cell_tissue; vital; 1.802 TPM; blood/CD8_STIM; dice_cd8_stim; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 12/133 (9.0%); Liver Hepatocellular Carcinoma (validation): 4/231 (1.7%); Lung Adenocarcinoma (validation): 1/105 (1.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Plays a key role in the replacement of histones to protamine in the elongating spermatids of mammals. In condensing spermatids, loaded onto the nucleosomes, where it promotes the recruitment and processing of protamines, which are responsible for histone eviction

Curated tissue note. Expressed by spermatids (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P09430 | HPA portal (live; not the pinned values)

TOMM20L | Unsafe / Protein

ENSG00000196860

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. duodenum / glandular cells: Low (Enhanced); small intestine / glandular cells: Low (Enhanced)

Cancer RNA. Kidney Chromophobe (TCGA): 2/64 (3.1%); Kidney Renal Papillary Cell Carcinoma (TCGA): 4/282 (1.4%); Pancreatic Adenocarcinoma (validation): 1/80 (1.2%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6UXN7 | HPA portal (live; not the pinned values)

TOP2A | Unsafe / Protein

ENSG00000131747

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bone marrow / hematopoietic cells: High (Enhanced); bronchus / respiratory epithelial cells: Low (Enhanced); colon / glandular cells: Medium (Enhanced); duodenum / glandular cells: Medium (Enhanced); esophagus / squamous epithelial cells: Medium ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 283/283 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); lymphoma: 12/12 (100.0%)

Biology. Key decatenating enzyme that alters DNA topology by binding to two double-stranded DNA molecules, generating a double-stranded break in one of the strands, passing the intact strand through the broken strand, and religating the broken strand (PubMed:17567603, PubMed:18790802, PubMed:22013166, PubMed:22323612, PubMed:27754753). May play a role in regulating the period length of BMAL1 transcriptional oscillation (By similarity)

Curated tissue note. Expressed in the tonsil, spleen, lymph node, thymus, skin, pancreas, testis, colon, kidney, liver, brain and lung (PubMed:9155056). Also found in high-grade lymphomas, squamous cell lung tumors and seminomas ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P11388 | HPA portal (live; not the pinned values)

TPK1 | Unsafe / Protein

ENSG00000196511

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum ... [full text in table]

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 93/100 (93.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 255/282 (90.4%); Breast Invasive Carcinoma (validation): 45/50 (90.0%)

Cancer IHC. prostate cancer: 11/11 (100.0%); breast cancer: 10/11 (90.9%)

Biology. Catalyzes the phosphorylation of thiamine to thiamine pyrophosphate (TPP) utilizing UTP and therefore links the biosynthesis of TPP to pyrimidines metabolism (PubMed:38547260). By producing thiamine pyrophosphate, a cofactor of the mitochondrial pyruvate dehydrogenase indirectly regulates pyruvate oxidation and lipogenesis (PubMed:38547260). Although it can also catalyze thiamine phosphorylation using ATP and CTP in vitro, it does so ... [full text in table]

Curated tissue note. Detected in heart, kidney, testis, small intestine and peripheral blood leukocytes, and at very low levels in a variety of tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H3S4 | HPA portal (live; not the pinned values)

TPPP2 | Unsafe / Protein

ENSG00000179636

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Enhanced); nasopharynx / respiratory epithelial cells: Medium (Enhanced)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 26/231 (11.3%); Liver Hepatocellular Carcinoma (TCGA): 19/362 (5.2%); Kidney Chromophobe (TCGA): 2/64 (3.1%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/9 (0.0%)

Biology. Probable regulator of microtubule dynamics required for sperm motility (Probable). In contrast to other members of the family, has no microtubule bundling activity (PubMed:17105200)

Curated tissue note. Expressed in spermatids (PubMed:23436708). Detected in liver cancer (at protein level) (PubMed:23436708)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P59282 | HPA portal (live; not the pinned values)

TPX2 | Unsafe / Protein

ENSG00000088325

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Enhanced); bone marrow / hematopoietic cells: High (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in molecular layer: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 283/283 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. breast cancer: 11/11 (100.0%); cervical cancer: 11/11 (100.0%)

Biology. Spindle assembly factor required for normal assembly of mitotic spindles. Required for normal assembly of microtubules during apoptosis. Required for chromatin and/or kinetochore dependent microtubule nucleation. Mediates AURKA localization to spindle microtubules (PubMed:18663142, PubMed:19208764, PubMed:37728657). Activates AURKA by promoting its autophosphorylation at 'Thr-288' and protects this residue against dephosphorylation ... [full text in table]

Curated tissue note. Expressed in lung carcinoma cell lines but not in normal lung tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9ULW0 | HPA portal (live; not the pinned values)

TRAPPC5 | Unsafe / Protein

ENSG00000181029

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); cerebral cortex / neuronal cells: Medium (Uncertain); colon / glandular cells: Low (Uncertain); duodenum / glandular cells: Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. colorectal cancer: 10/11 (90.9%); endometrial cancer: 10/12 (83.3%)

Biology. May play a role in vesicular transport from endoplasmic reticulum to Golgi

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IUR0 | HPA portal (live; not the pinned values)

TRAPPC9 | Unsafe / RNA

ENSG00000167632

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 13 nTPM (limit 2); amygdala: 11.5 nTPM (limit 2); basal ganglia: 13.6 nTPM (limit 2); bone marrow: 11 nTPM (limit 2); cerebellum: 15.8 nTPM (limit 2); cerebral cortex: 18.7 nTPM (limit 2); choroid plexus: 12.1 nTPM (limit 2); colon: 10.8 nTPM (limit 6); duodenum: 7.8 nTPM (limit 2); ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Functions as an activator of NF-kappa-B through increased phosphorylation of the IKK complex. May function in neuronal cells differentiation. May play a role in vesicular transport from endoplasmic reticulum to Golgi

Curated tissue note. Expressed at high levels in muscle and kidney and to a lower extent in brain, heart and placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96Q05 | HPA portal (live; not the pinned values)

TRDMT1 | Unsafe / Protein

ENSG00000107614

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 54/58 (93.1%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Colon Adenocarcinoma (validation): 277/486 (57.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); endometrial cancer: 12/12 (100.0%)

Biology. Specifically methylates cytosine 38 in the anticodon loop of tRNA(Asp) (PubMed:16424344). Has higher activity on tRNA(Asp) modified with queuosine at position 34 (PubMed:30093495)

Curated tissue note. Ubiquitous. Higher expression in testis, ovary and thymus and at much lower levels in spleen, prostate, colon, small intestine, and peripheral blood leukocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14717 | HPA portal (live; not the pinned values)

TRIM16 | Unsafe / Protein

ENSG00000221926

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); esophagus / squamous epithelial cells: High (Uncertain); kidney / cells in tubules: Medium (Uncertain); oral mucosa / squamous epithelial cells: Low (Uncertain); parathyroid gland / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. E3 ubiquitin-protein ligase that plays an essential role in the organization of autophagic response and ubiquitination upon lysosomal and phagosomal damages (PubMed:22629402, PubMed:27693506). Plays a role in the stress-induced biogenesis and degradation of protein aggresomes by regulating the p62-KEAP1-NRF2 signaling and particularly by modulating the ubiquitination levels and thus stability of NRF2 (PubMed:30143514). Acts as a ... [full text in table]

Curated tissue note. Highest levels found in testis, ovary, small intestine, colon, placenta, heart, skeletal muscle and mammary gland. More highly expressed in the fetus than in the corresponding adult tissues. Expressed in basal ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95361 | HPA portal (live; not the pinned values)

TRIM17 | Unsafe / Protein

ENSG00000162931

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / neuronal cells: Low (Approved); cerebral cortex / neuropil: Medium ... [full text in table]

Cancer RNA. Ovary Serous Cystadenocarcinoma (validation): 51/81 (63.0%); Breast Invasive Carcinoma (validation): 27/50 (54.0%); Ovary Serous Cystadenocarcinoma (TCGA): 145/349 (41.5%)

Cancer IHC. prostate cancer: 9/9 (100.0%); breast cancer: 10/11 (90.9%)

Biology. E3 ubiquitin ligase that plays important roles in the regulation of neuronal apoptosis, selective autophagy or cell proliferation (PubMed:19358823, PubMed:22023800, PubMed:27562068). Stimulates the degradation of kinetochore ZW10 interacting protein ZWINT in a proteasome-dependent manner, leading to negative regulation of cell proliferation (PubMed:22023800). Inhibits autophagic degradation of diverse known targets while contributing ... [full text in table]

Curated tissue note. Almost exclusively in the testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y577 | HPA portal (live; not the pinned values)

TRIM36 | Unsafe / RNA

ENSG00000152503

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 10.9 nTPM (limit 2); basal ganglia: 4.7 nTPM (limit 2); cerebral cortex: 15.8 nTPM (limit 2); choroid plexus: 14.4 nTPM (limit 2); colon: 7.2 nTPM (limit 6); duodenum: 13.7 nTPM (limit 2); hippocampal formation: 13 nTPM (limit 2); hypothalamus: 13.1 nTPM (limit 2); midbrain: 12.2 nTPM ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 476/480 (99.2%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Glioblastoma Multiforme (TCGA): 118/141 (83.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. E3 ubiquitin-protein ligase which mediates ubiquitination and subsequent proteasomal degradation of target proteins including CTNNB1, FOXA2 and AKT1 (PubMed:35768649, PubMed:37875418, PubMed:41323265). Involved in chromosome segregation and cell cycle regulation (PubMed:28087737). May play a role in the acrosome reaction and fertilization. Inhibits lung fibroblast proliferation, migration, and differentiation. Mechanistically, mediates ... [full text in table]

Curated tissue note. Highly expressed in testis, prostate and brain (PubMed:15145053). Weakly expressed in kidney, lung and heart (PubMed:15145053). Expressed in fetal tissues (PubMed:28087737)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NQ86 | HPA portal (live; not the pinned values)

TRIM40 | Unsafe / RNA

ENSG00000204614

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. small intestine: 2.9 nTPM (limit 2); small intestine: 2.9 nTPM (baseline 2)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 6/231 (2.6%); Colon Adenocarcinoma (validation): 6/486 (1.2%); Rectum Adenocarcinoma (validation): 2/207 (1.0%)

Cancer IHC. testis cancer: 9/12 (75.0%); prostate cancer: 8/12 (66.7%)

Biology. E3 ubiquitin-protein ligase that plays a role in the limitation of the innate immune response (PubMed:21474709, PubMed:29117565). Mediates inhibition of the RLR signaling pathway by ubiquitinating RIGI and IFIH1 receptors, leading to their proteasomal degradation (PubMed:21474709). Also promotes the neddylation of IKBKG/NEMO, stabilizing NFKBIA, and thereby inhibiting of NF-kappa-B nuclear translocation and activation (PubMed:21474709)

Curated tissue note. Highly expressed in normal gastrointestinal epithelia but that is down-regulated in gastrointestinal carcinomas and chronic inflammatory lesions of the gastrointestinal tract

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6P9F5 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

TRIM49D2 | Unsafe / High RNA

ENSG00000233802

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 57.7 nTPM; blood/basophil; hpa_immune_cell_samples; vital; 381 TPM; blood/basophil; hpa_immune_monaco; vital

Cancer RNA. Prostate Adenocarcinoma (TCGA): 9/480 (1.9%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Ovary Serous Cystadenocarcinoma (TCGA): 1/349 (0.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt C9J1S8 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

TRIM59 | Unsafe / RNA

ENSG00000213186

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 5 nTPM (limit 2); basal ganglia: 6.4 nTPM (limit 2); bone marrow: 17 nTPM (limit 2); cerebral cortex: 10.4 nTPM (limit 2); duodenum: 2.7 nTPM (limit 2); hippocampal formation: 8.8 nTPM (limit 2); hypothalamus: 4.2 nTPM (limit 2); lymph node: 11.8 nTPM (limit 6); midbrain: 9 nTPM (limit ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%); Rectum Adenocarcinoma (validation): 206/207 (99.5%)

Cancer IHC. prostate cancer: 12/12 (100.0%); urothelial cancer: 9/11 (81.8%)

Biology. E3 ubiquitin ligase involved in different processes such as development and immune response (PubMed:22588174, PubMed:30231667). Serves as a negative regulator for innate immune signaling pathways by suppressing RLR-induced activation of IRF3/7 and NF-kappa-B via interaction with adapter ECSIT (PubMed:22588174). Regulates autophagy through modulating both the transcription and the ubiquitination of BECN1 (PubMed:30231667). On the one ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IWR1 | HPA portal (live; not the pinned values)

TRIP13 | Unsafe / High RNA

ENSG00000071539

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 88.8 nTPM; lung/ciliated cells; hpa_single_cell_tissue; vital; 8.639 TPM; blood/CD8_STIM; dice_cd8_stim; vital

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Colon Adenocarcinoma (validation): 482/486 (99.2%); Lung Squamous Cell Carcinoma (validation): 67/68 (98.5%)

Cancer IHC. prostate cancer: 2/12 (16.7%); breast cancer: 1/12 (8.3%)

Biology. Plays a key role in chromosome recombination and chromosome structure development during meiosis. Required at early steps in meiotic recombination that leads to non-crossovers pathways. Also needed for efficient completion of homologous synapsis by influencing crossover distribution along the chromosomes affecting both crossovers and non-crossovers pathways. Also required for development of higher-order chromosome structures and is ... [full text in table]

Reconsideration. Resolve the documented non-permitted cell expression before any reproductive-restricted designation.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15645 | HPA portal (live; not the pinned values)

TRIQK | Unsafe / RNA

ENSG00000205133

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 12.3 nTPM (limit 10); adrenal gland: 21.2 nTPM (limit 2); amygdala: 12.3 nTPM (limit 2); basal ganglia: 14.8 nTPM (limit 2); bone marrow: 14.3 nTPM (limit 2); cerebellum: 15.7 nTPM (limit 2); cerebral cortex: 17.6 nTPM (limit 2); choroid plexus: 8.1 nTPM (limit 2); colon: 20 nTPM ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 100/100 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May play a role in cell growth and maintenance of cell morphology

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q629K1 | HPA portal (live; not the pinned values)

TRMT10A | Unsafe / RNA

ENSG00000145331

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.6 nTPM (limit 2); amygdala: 2.2 nTPM (limit 2); basal ganglia: 2.6 nTPM (limit 2); bone marrow: 2.2 nTPM (limit 2); cerebral cortex: 3.1 nTPM (limit 2); duodenum: 4.2 nTPM (limit 2); heart muscle: 2.2 nTPM (limit 2); hippocampal formation: 2.3 nTPM (limit 2); hypothalamus: 2.3 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Adenocarcinoma (validation): 75/105 (71.4%); Pancreatic Adenocarcinoma (validation): 49/80 (61.3%)

Cancer IHC. pancreatic cancer: 5/11 (45.5%); colorectal cancer: 5/12 (41.7%)

Biology. S-adenosyl-L-methionine-dependent guanine N(1)-methyltransferase that catalyzes the formation of N(1)-methylguanine at position 9 (m1G9) in tRNAs (PubMed:23042678, PubMed:25053765). Probably not able to catalyze formation of N(1)-methyladenine at position 9 (m1A9) in tRNAs (PubMed:23042678)

Curated tissue note. Expressed in embryonic and fetal brain. It is expressed throughout the dorsal telencephalon at 8 and 11 weeks of gestation, with highest expression in ventricular zone and marginal zone. Detected in cerebellar ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TBZ6 | HPA portal (live; not the pinned values)

TROAP | Unsafe / Protein

ENSG00000135451

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / cells in ... [full text in table]

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 283/283 (100.0%); Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%); Rectum Adenocarcinoma (TCGA): 88/88 (100.0%)

Cancer IHC. ovarian cancer: 9/12 (75.0%); urothelial cancer: 6/11 (54.5%)

Biology. Could be involved with bystin and trophinin in a cell adhesion molecule complex that mediates an initial attachment of the blastocyst to uterine epithelial cells at the time of the embryo implantation

Curated tissue note. Strong expression at implantation sites. Was exclusively localized to the apical side of the syncytiotrophoblast. Also found in macrophages

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q12815 | HPA portal (live; not the pinned values)

TRPM1 | Unsafe / Protein

ENSG00000134160

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. retina / inner nuclear layer: Medium (Enhanced); retina / outer plexiform layer: Medium (Enhanced)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 78/99 (78.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/10 (0.0%)

Biology. Constitutively open nonselective divalent cation-conducting channels which mediate the influx of Ca(2+), Mg(2+), Mn(2+), Ba(2+), and Ni(2+) into the cytoplasm, leading to membrane depolarization (PubMed:11535825, PubMed:19436059, PubMed:21278253). Impermeable to zinc ions (PubMed:21278253). In addition, forms heteromultimeric ion channels with TRPM3 which are permeable for calcium and zinc ions (PubMed:21278253). Plays an essential ... [full text in table]

Curated tissue note. Expressed in the retina where it localizes to the outer plexiform layer. Specifically, it is expressed in retinal bipolar cells (BPCs) of the ON subtype (PubMed:30027108). Highly expressed in benign ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z4N2 | HPA portal (live; not the pinned values)

TRPM3 | Unsafe / Protein

ENSG00000083067

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. kidney / proximal tubules (cell body): High (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 28/58 (48.3%); Kidney Renal Clear Cell Carcinoma (validation): 39/100 (39.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 65/282 (23.0%)

Cancer IHC. liver cancer: 1/10 (10.0%); prostate cancer: 1/12 (8.3%)

Biology. Constitutively active, non-selective divalent cation-conducting channel that is permeable to Ca(2+), Mn(2+), and Mg(2+), with a high permeability for Ca(2+). However, can be enhanced by increasing temperature and by ligands, including the endogenous neurosteroid pregnenolone sulfate and sphingosine-1 and suppressed by intracellular Mg(2+) (PubMed:12672799, PubMed:12672827, PubMed:32343227). Implicated in a variety of cellular ... [full text in table]

Curated tissue note. Expressed primarily in the kidney and, at lower levels, in brain, testis, ovary, pancreas and spinal cord. Expression in the brain and kidney was determined at protein level. In the kidney, expressed ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HCF6 | HPA portal (live; not the pinned values)

TRPV1 | Unsafe / RNA

ENSG00000196689

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.8 nTPM (limit 2); amygdala: 6.2 nTPM (limit 2); basal ganglia: 6.2 nTPM (limit 2); cerebellum: 13 nTPM (limit 2); cerebral cortex: 6.4 nTPM (limit 2); duodenum: 7.1 nTPM (limit 2); heart muscle: 7.3 nTPM (limit 2); hippocampal formation: 6.9 nTPM (limit 2); hypothalamus: 6.1 nTPM ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Liver Hepatocellular Carcinoma (validation): 195/231 (84.4%); Testicular Germ Cell Tumor (TCGA): 92/133 (69.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Non-selective calcium permeant cation channel involved in detection of noxious chemical and thermal stimuli (PubMed:11050376, PubMed:11243859, PubMed:11226139, PubMed:12077606). Seems to mediate proton influx and may be involved in intracellular acidosis in nociceptive neurons. Involved in mediation of inflammatory pain and hyperalgesia. Sensitized by a phosphatidylinositol second messenger system activated by receptor tyrosine ... [full text in table]

Curated tissue note. Widely expressed at low levels. Expression is elevated in dorsal root ganglia. In skin, expressed in cutaneous sensory nerve fibers, mast cells, epidermal keratinocytes, dermal blood vessels, the inner root ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NER1 | HPA portal (live; not the pinned values)

TSACC | Unsafe / RNA

ENSG00000163467

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Boundary. Failed bulk numeric comparisons are within 20% of their cutoffs; not an automatic rescue

Normal RNA / IHC. retina: 2.2 nTPM (limit 2); retina: 2.2 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 87/133 (65.4%); Rectum Adenocarcinoma (validation): 91/207 (44.0%); Colon Adenocarcinoma (validation): 181/486 (37.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Co-chaperone that facilitates HSP-mediated activation of TSSK6

Curated tissue note. Expressed in testis but is absent from mature sperm

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96A04 | HPA portal (live; not the pinned values)

TSGA10 | Unsafe / RNA

ENSG00000135951

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3.1 nTPM (limit 2); kidney: 3 nTPM (limit 2); pituitary gland: 2.4 nTPM (limit 2); retina: 2.5 nTPM (limit 2); choroid plexus: 3.1 nTPM (baseline 2); kidney: 3 nTPM (baseline 2); pituitary gland: 2.4 nTPM (baseline 2); retina: 2.5 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 44/50 (88.0%); Glioblastoma Multiforme (validation): 39/58 (67.2%); Pancreatic Adenocarcinoma (validation): 52/80 (65.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays a role in spermatogenesis (PubMed:28905369). When overexpressed, prevents nuclear localization of HIF1A (By similarity)

Curated tissue note. Expressed in the testis, in spermatozoa (at protein level) (PubMed:11179690, PubMed:28905369). Expressed in actively dividing fetal tissues, including sternum, intestine, limb, kidney and stomach ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BZW7 | HPA portal (live; not the pinned values)

TSPAN16 | Unsafe / Protein

ENSG00000130167

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); kidney / cells in tubules: Medium (Enhanced); stomach 1 / glandular cells: Low (Enhanced); stomach 2 / glandular cells: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%); Glioblastoma Multiforme (validation): 1/58 (1.7%); Uterine Corpus Endometrial Carcinoma (TCGA): 2/176 (1.1%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Broadly expressed in most human tissues and cell lines including neural and bone marrow derived tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UKR8 | HPA portal (live; not the pinned values)

TSPY9 | Unsafe / Protein

ENSG00000238074

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Supported); stomach 1 / glandular cells: Low (Supported); stomach 2 / glandular cells: Low (Supported)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 33/133 (24.8%); Lung Squamous Cell Carcinoma (validation): 3/68 (4.4%); Lung Squamous Cell Carcinoma (TCGA): 20/489 (4.1%)

Cancer IHC. testis cancer: 3/12 (25.0%); breast cancer: 0/10 (0.0%)

Biology. May be involved in sperm differentiation and proliferation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A0A494C1R9 | HPA portal (live; not the pinned values)

TSSK3 | Unsafe / RNA

ENSG00000162526

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.6 nTPM (limit 2); bone marrow: 12.5 nTPM (limit 2); cerebellum: 5.4 nTPM (limit 2); duodenum: 3.3 nTPM (limit 2); liver: 2.6 nTPM (limit 2); lung: 2.6 nTPM (limit 2); pancreas: 2.1 nTPM (limit 2); retina: 4.2 nTPM (limit 2); skeletal muscle: 2.9 nTPM (limit 2); small intestine: 3.9 ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 42/50 (84.0%); Liver Hepatocellular Carcinoma (validation): 146/231 (63.2%); Rectum Adenocarcinoma (validation): 50/207 (24.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Serine/threonine protein kinase required for spermatid development and male fertility

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96PN8 | HPA portal (live; not the pinned values)

TTC16 | Unsafe / High RNA

ENSG00000167094

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 65.6 nTPM; pbmc/t-cells; hpa_single_cell_tissue; vital; 82.56 TPM; blood/TH1; dice_th1; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 8/50 (16.0%); Lung Adenocarcinoma (TCGA): 6/497 (1.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 2/176 (1.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEE8 | HPA portal (live; not the pinned values)

TTC21A | Unsafe / High RNA

ENSG00000168026

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 107 nTPM; bronchus/ciliated cells; hpa_single_cell_tissue; vital; 58.2 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Breast Invasive Carcinoma (validation): 47/50 (94.0%); Glioblastoma Multiforme (validation): 33/58 (56.9%); Pancreatic Adenocarcinoma (validation): 20/80 (25.0%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Intraflagellar transport (IFT)-associated protein required for spermatogenesis (PubMed:30929735). Required for sperm flagellar formation and intraflagellar transport (PubMed:30929735)

Curated tissue note. Strongly expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NDW8 | HPA portal (live; not the pinned values)

TTC29 | Unsafe / Protein

ENSG00000137473

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Uncertain); duodenum / glandular cells: Low (Uncertain); heart muscle / cardiomyocytes: Low (Uncertain); kidney / cells in tubules: Medium (Uncertain); nasopharynx / respiratory epithelial cells: Low (Uncertain); rectum / glandular cells: Low ... [full text in table]

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 17/176 (9.7%); Breast Invasive Carcinoma (validation): 4/50 (8.0%); Glioblastoma Multiforme (validation): 4/58 (6.9%)

Cancer IHC. stomach cancer: 1/12 (8.3%); breast cancer: 0/10 (0.0%)

Biology. Axonemal protein which is implicated in axonemal and/or peri-axonemal structure assembly and regulates flagellum assembly and beating and therefore sperm motility

Curated tissue note. Expressed in spermatozoa (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NA56 | HPA portal (live; not the pinned values)

TTK | Unsafe / Protein

ENSG00000112742

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. 62.3 nTPM; tongue/basal keratinocytes; hpa_single_cell_tissue; protected; 4.674 TPM; blood/TREG_MEM; dice_treg_mem; vital

Cancer RNA. Colon Adenocarcinoma (validation): 483/486 (99.4%); Lung Squamous Cell Carcinoma (validation): 67/68 (98.5%); Rectum Adenocarcinoma (validation): 201/207 (97.1%)

Cancer IHC. testis cancer: 11/11 (100.0%); skin cancer: 11/12 (91.7%)

Biology. Involved in mitotic spindle assembly checkpoint signaling, a process that delays anaphase until chromosomes are bioriented on the spindle, and in the repair of incorrect mitotic kinetochore-spindle microtubule attachments (PubMed:18243099, PubMed:28441529, PubMed:29162720). Phosphorylates MAD1L1 to promote the mitotic spindle assembly checkpoint (PubMed:18243099, PubMed:29162720). Phosphorylates CDCA8/Borealin leading to enhanced AURKB ... [full text in table]

Curated tissue note. Present in rapidly proliferating cell lines

Therapeutic evidence. Trial registration only.

UniProt P33981 | Trial/source record | HPA portal (live; not the pinned values)

TTLL10 | Unsafe / RNA

ENSG00000162571

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.9 nTPM (limit 2); lung: 2.7 nTPM (limit 2); choroid plexus: 2.9 nTPM (baseline 2); lung: 2.7 nTPM (baseline 2)

Cancer RNA. Uterine Corpus Endometrial Carcinoma (TCGA): 24/176 (13.6%); Pancreatic Adenocarcinoma (validation): 7/80 (8.8%); Ovary Serous Cystadenocarcinoma (TCGA): 19/349 (5.4%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 11/12 (91.7%)

Biology. Inactive polyglycylase

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZVT0 | HPA portal (live; not the pinned values)

TTLL11 | Unsafe / Protein

ENSG00000175764

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 42/50 (84.0%); Rectum Adenocarcinoma (validation): 162/207 (78.3%)

Cancer IHC. renal cancer: 9/12 (75.0%); pancreatic cancer: 8/11 (72.7%)

Biology. Polyglutamylase which modifies tubulin, generating polyglutamate side chains of variable lengths on the gamma-carboxyl group of specific glutamate residues within the C-terminal tail of tubulin. Preferentially mediates ATP-dependent polyglutamate long side-chain elongation over the initiation step of the polyglutamylation reaction. Preferentially modifies the alpha-tubulin tail over a beta-tail (By similarity). Required for CCSAP ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NHH1 | HPA portal (live; not the pinned values)

TTLL6 | Unsafe / Protein

ENSG00000170703

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): High (Enhanced); bronchus / ciliated cells (tip of cilia): Low (Enhanced); nasopharynx / ciliated cells (ciliary rootlets): High (Enhanced)

Cancer RNA. Kidney Renal Clear Cell Carcinoma (validation): 17/100 (17.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 33/282 (11.7%); Glioblastoma Multiforme (validation): 5/58 (8.6%)

Cancer IHC. melanoma: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. Polyglutamylase which modifies both tubulin and non-tubulin proteins, generating alpha-linked polyglutamate side chains on the gamma-carboxyl group of specific glutamate residues of target proteins. Preferentially mediates ATP-dependent long polyglutamate chain elongation over the initiation step of the polyglutamylation reaction. Preferentially modifies the alpha-tubulin tail over a beta-tail. Promotes tubulin polyglutamylation which ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N841 | HPA portal (live; not the pinned values)

TTLL9 | Unsafe / RNA

ENSG00000131044

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.7 nTPM (limit 2); choroid plexus: 15 nTPM (limit 2); hippocampal formation: 2.5 nTPM (limit 2); hypothalamus: 2.6 nTPM (limit 2); spinal cord: 2.3 nTPM (limit 2); basal ganglia: 2.7 nTPM (baseline 2); choroid plexus: 15 nTPM (baseline 2); hippocampal formation: 2.5 nTPM (baseline ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 50/58 (86.2%); Breast Invasive Carcinoma (validation): 21/50 (42.0%); Ovary Serous Cystadenocarcinoma (validation): 9/81 (11.1%)

Cancer IHC. skin cancer: 7/11 (63.6%); testis cancer: 7/12 (58.3%)

Biology. Probable tubulin polyglutamylase that generates side chains of glutamate on the gamma-carboxyl group of specific glutamate residues within the C-terminal tail of target proteins. Similar to TTLL1, may acquire enzymatic activity only in complex with other proteins as it is most likely lacking domains important for autonomous activity. Mediates tubulin polyglutamylation which induces establishment of microtubule heterogeneity in sperm ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3SXZ7 | HPA portal (live; not the pinned values)

TUBA3D | Unsafe / RNA

ENSG00000075886

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. heart muscle: 83.4 nTPM (limit 2); heart muscle: 83.4 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 34/50 (68.0%); Prostate Adenocarcinoma (TCGA): 212/480 (44.2%); Testicular Germ Cell Tumor (TCGA): 56/133 (42.1%)

Cancer IHC. testis cancer: 7/11 (63.6%); glioma: 5/11 (45.5%)

Biology. Tubulin is the major constituent of microtubules, a cylinder consisting of laterally associated linear protofilaments composed of alpha- and beta-tubulin heterodimers. Microtubules grow by the addition of GTP-tubulin dimers to the microtubule end, where a stabilizing cap forms. Below the cap, tubulin dimers are in GDP-bound state, owing to GTPase activity of alpha-tubulin

Curated tissue note. Expressed in testis | Expressed in the cornea, sclera, and peripheral blood (PubMed:29051577)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P0DPH7 | UniProt P0DPH8 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

TUBA3E | Unsafe / RNA

ENSG00000152086

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. heart muscle: 51.5 nTPM (limit 2); heart muscle: 51.5 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 18/50 (36.0%); Testicular Germ Cell Tumor (TCGA): 21/133 (15.8%); Uterine Corpus Endometrial Carcinoma (TCGA): 17/176 (9.7%)

Cancer IHC. testis cancer: 7/11 (63.6%); glioma: 5/11 (45.5%)

Biology. Tubulin is the major constituent of microtubules, a cylinder consisting of laterally associated linear protofilaments composed of alpha- and beta-tubulin heterodimers. Microtubules grow by the addition of GTP-tubulin dimers to the microtubule end, where a stabilizing cap forms. Below the cap, tubulin dimers are in GDP-bound state, owing to GTPase activity of alpha-tubulin

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PEY2 | HPA portal (live; not the pinned values)

TUBB8B | Unsafe / Protein

ENSG00000173213

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 48/133 (36.1%); Uterine Corpus Endometrial Carcinoma (TCGA): 10/176 (5.7%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 9/283 (3.2%)

Cancer IHC. glioma: 2/9 (22.2%); pancreatic cancer: 1/10 (10.0%)

Biology. Tubulin is the major constituent of microtubules, a cylinder consisting of laterally associated linear protofilaments composed of alpha- and beta-tubulin heterodimers. Microtubules grow by the addition of GTP-tubulin dimers to the microtubule end, where a stabilizing cap forms. Below the cap, tubulin dimers are in GDP-bound state, owing to GTPase activity of alpha-tubulin

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NNZ2 | HPA portal (live; not the pinned values)

TUBGCP3 | Unsafe / Protein

ENSG00000126216

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); ovarian cancer: 12/12 (100.0%)

Biology. Component of the gamma-tubulin ring complex (gTuRC) which mediates microtubule nucleation (PubMed:38305685, PubMed:38609661, PubMed:39321809, PubMed:9566967). The gTuRC regulates the minus-end nucleation of alpha-beta tubulin heterodimers that grow into microtubule protafilaments, a critical step in centrosome duplication and spindle formation (PubMed:38305685, PubMed:38609661, PubMed:39321809)

Curated tissue note. Ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96CW5 | HPA portal (live; not the pinned values)

TULP2 | Unsafe / High RNA

ENSG00000104804

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 12 nTPM; esophagus/schwann cells; hpa_single_cell_tissue; protected; 43.49 TPM; blood/TH1; dice_th1; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 28/133 (21.1%); Rectum Adenocarcinoma (validation): 24/207 (11.6%); Colon Adenocarcinoma (validation): 16/486 (3.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Strongly expressed in testis. Also expressed in retina. Expressed in cancer cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00295 | HPA portal (live; not the pinned values)

TVP23C | Unsafe / Protein

ENSG00000175106

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); cerebellum / Purkinje cells: Medium (Uncertain); cerebral cortex / endothelial cells: High (Uncertain); colon / endothelial cells: High ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 69/80 (86.2%); Colon Adenocarcinoma (validation): 345/486 (71.0%); Rectum Adenocarcinoma (validation): 145/207 (70.0%)

Cancer IHC. prostate cancer: 10/11 (90.9%); melanoma: 3/8 (37.5%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96ET8 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

TXNDC2 | Unsafe / Protein

ENSG00000168454

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Enhanced); colon / glandular cells: Low (Enhanced); duodenum / glandular cells: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); lung / macrophages: Low (Enhanced); parathyroid gland / glandular cells: Low (Enhanced); rectum / glandular cells: ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Lung Squamous Cell Carcinoma (validation): 1/68 (1.5%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. liver cancer: 1/12 (8.3%); breast cancer: 0/12 (0.0%)

Biology. Probably plays a regulatory role in sperm development. May participate in regulation of fibrous sheath (FS) assembly by supporting the formation of disulfide bonds during sperm tail morphogenesis. May also be required to rectify incorrect disulfide pairing and generate suitable pairs between the FS constituents. Can reduce disulfide bonds in vitro in the presence of NADP and thioredoxin reductase

Curated tissue note. Testis-specific. Only expressed during spermiogenesis, prominently in round and elongating spermatids

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86VQ3 | HPA portal (live; not the pinned values)

UBE2S | Unsafe / Protein

ENSG00000108106

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. colon / endothelial cells: Low (Approved); lymph node / germinal center cells: Medium (Approved); nasopharynx / respiratory epithelial cells: Low (Approved); urinary bladder / urothelial cells: Low (Approved)

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%)

Cancer IHC. melanoma: 3/12 (25.0%); breast cancer: 1/12 (8.3%)

Biology. Accepts ubiquitin from the E1 complex and catalyzes its covalent attachment to other proteins (PubMed:19820702, PubMed:19822757, PubMed:22496338, PubMed:27259151). Catalyzes 'Lys-11'-linked polyubiquitination. Acts as an essential factor of the anaphase promoting complex/cyclosome (APC/C), a cell cycle-regulated ubiquitin ligase that controls progression through mitosis (PubMed:19820702, PubMed:19822757, PubMed:27259151, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q16763 | HPA portal (live; not the pinned values)

UBE2V1 | Unsafe / RNA

ENSG00000244687

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 59.1 nTPM (limit 10); adrenal gland: 54.4 nTPM (limit 2); amygdala: 61.1 nTPM (limit 2); basal ganglia: 53.6 nTPM (limit 2); bone marrow: 50.6 nTPM (limit 2); cerebellum: 53.1 nTPM (limit 2); cerebral cortex: 78.3 nTPM (limit 2); choroid plexus: 77 nTPM (limit 2); colon: 67.1 nTPM ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Has no ubiquitin ligase activity on its own. The UBE2V1-UBE2N heterodimer catalyzes the synthesis of non-canonical poly-ubiquitin chains that are linked through Lys-63. This type of poly-ubiquitination activates IKK and does not seem to involve protein degradation by the proteasome. Plays a role in the activation of NF-kappa-B mediated by IL1B, TNF, TRAF6 and TRAF2. Mediates transcriptional activation of target genes. Plays a role in ... [full text in table]

Curated tissue note. Highly expressed in thyroid, pancreas, spinal cord, lymph node, trachea, adrenal gland, bone marrow and pancreas. Detected at low levels in heart, breast, placenta, brain, liver, kidney, stomach and lung

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13404 | HPA portal (live; not the pinned values)

UBE3C | Unsafe / Protein

ENSG00000009335

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: High (Approved); bronchus / ciliated cells (cell body): Medium (Approved); bronchus / ciliated cells (ciliary rootlets): ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Stomach Adenocarcinoma (TCGA): 346/346 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. liver cancer: 12/12 (100.0%); stomach cancer: 10/10 (100.0%)

Biology. E3 ubiquitin-protein ligase that specifically catalyzes 'Lys-29'- and 'Lys-48'-linked polyubiquitin chains (PubMed:11278995, PubMed:12692129, PubMed:16341092, PubMed:16601690, PubMed:24158444, PubMed:24811749, PubMed:25752573, PubMed:25752577, PubMed:32039437, PubMed:33637724, PubMed:34239127). Accepts ubiquitin from the E2 ubiquitin-conjugating enzyme UBE2D1 in the form of a thioester and then directly transfers the ubiquitin to ... [full text in table]

Curated tissue note. Highly expressed in skeletal muscle. Detected at much lower levels in kidney and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15386 | HPA portal (live; not the pinned values)

UBE3D | Unsafe / Protein

ENSG00000118420

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 51/58 (87.9%); Pancreatic Adenocarcinoma (validation): 28/80 (35.0%); Rectum Adenocarcinoma (TCGA): 30/88 (34.1%)

Cancer IHC. liver cancer: 5/12 (41.7%); ovarian cancer: 3/12 (25.0%)

Biology. E3 ubiquitin-protein ligase which accepts ubiquitin from specific E2 ubiquitin-conjugating enzymes, and transfers it to substrates, generally promoting their degradation by the proteasome (PubMed:15749827). Independently of its E3 ubiquitin-protein ligase activity, acts as an inhibitor of CPSF3 endonuclease activity by blocking CPSF3 active site (PubMed:39032490)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z6J8 | HPA portal (live; not the pinned values)

UBL4B | Unsafe / RNA

ENSG00000186150

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 3.2 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (validation): 1/58 (1.7%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%); Glioblastoma Multiforme (TCGA): 1/141 (0.7%)

Cancer IHC. lung cancer: 3/12 (25.0%); melanoma: 3/12 (25.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N7F7 | HPA portal (live; not the pinned values)

UBXN10 | Unsafe / Protein

ENSG00000162543

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 50/50 (100.0%); Thyroid Carcinoma (TCGA): 344/495 (69.5%); Colon Adenocarcinoma (validation): 309/486 (63.6%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); melanoma: 10/11 (90.9%)

Biology. VCP/p97-binding protein required for ciliogenesis (PubMed:26389662). Acts as a tethering factor that facilitates recruitment of VCP/p97 to the intraflagellar transport complex B (IFT-B) in cilia (PubMed:26389662). UBX domain-containing proteins act as tethering factors for VCP/p97 and may specify substrate specificity of VCP/p97 (PubMed:26389662)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96LJ8 | HPA portal (live; not the pinned values)

UCN2 | Unsafe / RNA

ENSG00000145040

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 12.8 nTPM (limit 10); skin: 12.8 nTPM (baseline 10)

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 58/99 (58.6%); Head and Neck Squamous Cell Carcinoma (TCGA): 236/492 (48.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 109/283 (38.5%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Hormone that plays a central role in whole body adaptation to stress (PubMed:11329063). Released by the hypothalamus primarily in response to physical or psychological stress and acts by binding to CRH receptor CRHR2 (PubMed:20966082). UCN2-dependent signaling regulates cardiovascular function, promotes skeletal muscle hypertrophy and manage energy balance and acts as an anorexigenic agent (By similarity). In contrast to CRH and UCN, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96RP3 | HPA portal (live; not the pinned values)

UGGT2 | Unsafe / RNA

ENSG00000102595

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 11.4 nTPM (limit 10); adrenal gland: 6.7 nTPM (limit 2); amygdala: 4.3 nTPM (limit 2); basal ganglia: 3.7 nTPM (limit 2); bone marrow: 3.7 nTPM (limit 2); cerebral cortex: 5.3 nTPM (limit 2); choroid plexus: 6.9 nTPM (limit 2); colon: 6.6 nTPM (limit 6); duodenum: 5.2 nTPM (limit ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. endometrial cancer: 9/11 (81.8%); cervical cancer: 7/12 (58.3%)

Biology. Recognizes glycoproteins with minor folding defects. Reglucosylates single N-glycans near the misfolded part of the protein, thus providing quality control for protein folding in the endoplasmic reticulum. Reglucosylated proteins are recognized by calreticulin for recycling to the endoplasmic reticulum and refolding or degradation

Curated tissue note. Higher levels in kidney, pancreas, heart, and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NYU1 | HPA portal (live; not the pinned values)

ULBP3 | Unsafe / Protein

ENSG00000131019

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / neuronal cells: Low (Approved); esophagus / squamous epithelial cells: Low (Approved); lymph node / germinal center cells: Low (Approved); lymph node / non-germinal center cells: ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 378/495 (76.4%); Colon Adenocarcinoma (validation): 368/486 (75.7%); Glioblastoma Multiforme (validation): 40/58 (69.0%)

Cancer IHC. breast cancer: 1/11 (9.1%); cervical cancer: 0/12 (0.0%)

Biology. Binds and activates the KLRK1/NKG2D receptor, mediating natural killer cell cytotoxicity

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BZM4 | HPA portal (live; not the pinned values)

ULK2 | Unsafe / Protein

ENSG00000083290

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Low (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 48/50 (96.0%); Kidney Renal Clear Cell Carcinoma (validation): 87/100 (87.0%)

Cancer IHC. liver cancer: 12/12 (100.0%); ovarian cancer: 12/12 (100.0%)

Biology. Serine/threonine-protein kinase involved in autophagy in response to starvation. Acts upstream of phosphatidylinositol 3-kinase PIK3C3 to regulate the formation of autophagophores, the precursors of autophagosomes. Part of regulatory feedback loops in autophagy: acts both as a downstream effector and a negative regulator of mammalian target of rapamycin complex 1 (mTORC1) via interaction with RPTOR. Activated via phosphorylation by ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IYT8 | HPA portal (live; not the pinned values)

ULK4 | Unsafe / Protein

ENSG00000168038

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 42/50 (84.0%); Colon Adenocarcinoma (validation): 379/486 (78.0%)

Cancer IHC. melanoma: 9/10 (90.0%); colorectal cancer: 9/12 (75.0%)

Biology. Pseudokinase that acts as a molecular adapter in the smoothened signaling pathway (PubMed:38096226). Required to relieve inhibition of the GLI2 transcription factor in the presence of hedgehog (DHH, IHH or SHH) ligand: acts by mediating the association between STK36 and its ligand GLI2 in ciliary tip, promoting GLI2 phosphorylation and dissociation from SUFU inhibitor, followed by GLI2 translocation to the nucleus (PubMed:38096226)

Curated tissue note. Expressed in the brain, mainly in postmitotic neurons, including GABAergic neurons, but not in astrocytes (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96C45 | HPA portal (live; not the pinned values)

UQCR11 | Unsafe / RNA

ENSG00000127540

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 68.8 nTPM (limit 10); adrenal gland: 77.7 nTPM (limit 2); amygdala: 139.8 nTPM (limit 2); basal ganglia: 112.7 nTPM (limit 2); bone marrow: 39 nTPM (limit 2); cerebellum: 47.2 nTPM (limit 2); cerebral cortex: 106.3 nTPM (limit 2); choroid plexus: 95.4 nTPM (limit 2); colon: 88.6 ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Component of the ubiquinol-cytochrome c oxidoreductase, a multisubunit transmembrane complex that is part of the mitochondrial electron transport chain which drives oxidative phosphorylation. The respiratory chain contains 3 multisubunit complexes succinate dehydrogenase (complex II, CII), ubiquinol-cytochrome c oxidoreductase (cytochrome b-c1 complex, complex III, CIII) and cytochrome c oxidase (complex IV, CIV), that cooperate to ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O14957 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

USH2A | Unsafe / RNA

ENSG00000042781

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. liver: 6.7 nTPM (limit 2); retina: 18.6 nTPM (limit 2); liver: 6.7 nTPM (baseline 2); retina: 18.6 nTPM (baseline 2)

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 14/231 (6.1%); Liver Hepatocellular Carcinoma (TCGA): 5/362 (1.4%); Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Involved in hearing and vision as member of the USH2 complex. In the inner ear, required for the maintenance of the hair bundle ankle formation, which connects growing stereocilia in developing cochlear hair cells. In retina photoreceptors, the USH2 complex is required for the maintenance of periciliary membrane complex that seems to play a role in regulating intracellular protein transport

Curated tissue note. Present in the basement membrane of many, but not all tissues. Expressed in retina, cochlea, small and large intestine, pancreas, bladder, prostate, esophagus, trachea, thymus, salivary glands, placenta, ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75445 | HPA portal (live; not the pinned values)

USP25 | Unsafe / Protein

ENSG00000155313

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. endometrial cancer: 11/11 (100.0%); lymphoma: 11/11 (100.0%)

Biology. Deubiquitinating enzyme that hydrolyzes ubiquitin moieties conjugated to substrates and thus, functions in various biological processes including inflammation and immune response (PubMed:29518389, PubMed:37683630). Modulates the Wnt/beta-catenin pathway by deubiquitinating and stabilizing tankyrases TNKS1 and TNKS2 (PubMed:28619731, PubMed:30926243, PubMed:38875478). Regulates KEAP1-NRF2 axis in the defense against oxidative assaults ... [full text in table]

Curated tissue note. Isoform USP25a is found in most adult and fetal tissues; expression is moderately high in testis, pancreas, kidney, skeletal muscle, liver, lung, placenta, heart, but very low in peripheral blood, colon, small ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UHP3 | HPA portal (live; not the pinned values)

USP37 | Unsafe / Protein

ENSG00000135913

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. colorectal cancer: 4/11 (36.4%); ovarian cancer: 3/11 (27.3%)

Biology. Deubiquitinase that plays a role in different processes including cell cycle regulation, DNA replication or DNA damage response (PubMed:26299517, PubMed:27296872, PubMed:31911859, PubMed:34509474). Antagonizes the anaphase-promoting complex (APC/C) during G1/S transition by mediating deubiquitination of cyclin-A (CCNA1 and CCNA2), thereby promoting S phase entry. Specifically mediates deubiquitination of 'Lys-11'-linked polyubiquitin ... [full text in table]

Curated tissue note. Expressed in brain and prostate

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86T82 | HPA portal (live; not the pinned values)

USP44 | Unsafe / RNA

ENSG00000136014

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 3.9 nTPM (limit 2); lung: 3.7 nTPM (limit 2); pancreas: 2.3 nTPM (limit 2); cerebellum: 3.9 nTPM (baseline 2); lung: 3.7 nTPM (baseline 2); pancreas: 2.3 nTPM (baseline 2)

Cancer RNA. Lung Adenocarcinoma (validation): 32/105 (30.5%); Testicular Germ Cell Tumor (TCGA): 39/133 (29.3%); Lung Adenocarcinoma (TCGA): 51/497 (10.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Deubiquitinase that plays a key regulatory role in the spindle assembly checkpoint or mitotic checkpoint by preventing premature anaphase onset. Acts by specifically mediating deubiquitination of CDC20, a negative regulator of the anaphase promoting complex/cyclosome (APC/C) (PubMed:17443180). Deubiquitination of CDC20 leads to stabilize the MAD2L1-CDC20-APC/C ternary complex (also named mitotic checkpoint complex), thereby preventing ... [full text in table]

Curated tissue note. Expressed in testis. Expressed at high levels in T-cell acute lymphoblastic leukemia

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0E7 | HPA portal (live; not the pinned values)

USP49 | Unsafe / Protein

ENSG00000164663

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: High ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 51/58 (87.9%); Rectum Adenocarcinoma (validation): 129/207 (62.3%); Colon Adenocarcinoma (validation): 267/486 (54.9%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); breast cancer: 11/12 (91.7%)

Biology. Deubiquitinase that plays a role in several cellular processes including transcriptional regulation, cell cycle progression or innate immunity. Specifically deubiquitinates histone H2B at 'Lys-120' (H2BK120Ub), a specific tag for epigenetic transcriptional activation and acts as a regulator of mRNA splicing (PubMed:23824326). Antagonizes DNA double-strand break-induced ubiquitination of H2AX thereby participating in the maintenance of ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q70CQ1 | HPA portal (live; not the pinned values)

USP51 | Unsafe / Protein

ENSG00000247746

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 45/58 (77.6%); Breast Invasive Carcinoma (validation): 34/50 (68.0%); Kidney Chromophobe (TCGA): 30/64 (46.9%)

Cancer IHC. breast cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. Specifically deubiquitinates 'Lys-14' (H2AK13Ub) and 'Lys-16'(H2AK15Ub) of histone H2A regulating the DNA damage response at double-strand breaks (DSBs) (PubMed:27083998, PubMed:33022275). USP51 is recruited to chromatin after DNA damage and regulates the dynamic assembly/disassembly of TP53BP1 and BRCA1. Functions in DNA double-strand break repair also by mediating the deubiquitination and subsequent stabilization of DGCR8, leading to ... [full text in table]

Curated tissue note. Expressed in prostate, brain, lung, aorta and kidney

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q70EK9 | HPA portal (live; not the pinned values)

UTF1 | Unsafe / High RNA

ENSG00000171794

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 66.6 nTPM; adipose tissue/macrophages; hpa_single_cell_tissue; vital; 1.703 TPM; blood/MONOCYTES; dice_monocytes; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 117/133 (88.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 2/176 (1.1%); Rectum Adenocarcinoma (validation): 1/207 (0.5%)

Cancer IHC. testis cancer: 8/11 (72.7%); breast cancer: 0/11 (0.0%)

Biology. Acts as a transcriptional coactivator of ATF2

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5T230 | HPA portal (live; not the pinned values)

VAMP2 | Unsafe / RNA

ENSG00000220205

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 85.7 nTPM (limit 10); adrenal gland: 122.7 nTPM (limit 2); amygdala: 326.1 nTPM (limit 2); basal ganglia: 296.4 nTPM (limit 2); bone marrow: 170.1 nTPM (limit 2); cerebellum: 377.7 nTPM (limit 2); cerebral cortex: 470.8 nTPM (limit 2); choroid plexus: 75 nTPM (limit 2); colon: 92.3 ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. SNAREs (soluble N-ethylmaleimide-sensitive factor-attachment protein receptors) are essential proteins for intracellular membrane fusion. SNAREs localized on opposing membranes assemble to form a trans-SNARE complex, an extended, parallel four-helix bundle whose assembly releases energy that drives membrane fusion. The core SNARE complex typically consists of four alpha-helical domains, three from target membrane SNAREs (t-SNAREs) and ... [full text in table]

Curated tissue note. Nervous system and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P63027 | HPA portal (live; not the pinned values)

VASH2 | Unsafe / RNA

ENSG00000143494

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pituitary gland: 2.5 nTPM (limit 2); pituitary gland: 2.5 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 38/50 (76.0%); Testicular Germ Cell Tumor (TCGA): 86/133 (64.7%); Glioblastoma Multiforme (validation): 17/58 (29.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Tyrosine carboxypeptidase that removes the C-terminal tyrosine residue of alpha-tubulin, thereby regulating microtubule dynamics and function (PubMed:29146869). Critical for spindle function and accurate chromosome segregation during mitosis since microtubule detyrosination regulates mitotic spindle length and postioning (PubMed:31171830). Acts as an activator of angiogenesis: expressed in infiltrating mononuclear cells in the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86V25 | HPA portal (live; not the pinned values)

VAV3 | Unsafe / Protein

ENSG00000134215

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 50/50 (100.0%); Rectum Adenocarcinoma (validation): 203/207 (98.1%); Kidney Renal Papillary Cell Carcinoma (TCGA): 276/282 (97.9%)

Cancer IHC. colorectal cancer: 7/12 (58.3%); pancreatic cancer: 6/12 (50.0%)

Biology. Exchange factor for GTP-binding proteins RhoA, RhoG and, to a lesser extent, Rac1. Binds physically to the nucleotide-free states of those GTPases. Plays an important role in angiogenesis. Its recruitment by phosphorylated EPHA2 is critical for EFNA1-induced RAC1 GTPase activation and vascular endothelial cell migration and assembly (By similarity). May be important for integrin-mediated signaling, at least in some cell types. In ... [full text in table]

Curated tissue note. Isoform 1 and isoform 3 are widely expressed; both are expressed at very low levels in skeletal muscle. In keratinocytes, isoform 1 is less abundant than isoform 3. Isoform 3 is detected at very low levels, if ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UKW4 | HPA portal (live; not the pinned values)

VCPIP1 | Unsafe / RNA

ENSG00000175073

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.5 nTPM (limit 2); amygdala: 2.8 nTPM (limit 2); basal ganglia: 2.9 nTPM (limit 2); bone marrow: 20 nTPM (limit 2); cerebellum: 2.9 nTPM (limit 2); cerebral cortex: 5.7 nTPM (limit 2); duodenum: 3.7 nTPM (limit 2); heart muscle: 2.8 nTPM (limit 2); hippocampal formation: 3 nTPM ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. endometrial cancer: 2/12 (16.7%); ovarian cancer: 2/12 (16.7%)

Biology. Deubiquitinating enzyme involved in DNA repair and reassembly of the Golgi apparatus and the endoplasmic reticulum following mitosis (PubMed:32649882). Necessary for VCP-mediated reassembly of Golgi stacks after mitosis (By similarity). Plays a role in VCP-mediated formation of transitional endoplasmic reticulum (tER) (By similarity). Mediates dissociation of the ternary complex containing STX5A, NSFL1C and VCP (By similarity). Also ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96JH7 | HPA portal (live; not the pinned values)

VCX2 | Unsafe / Protein

ENSG00000177504

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 7/133 (5.3%); Skin Cuteneous Melanoma (TCGA): 2/99 (2.0%); Stomach Adenocarcinoma (TCGA): 5/346 (1.4%)

Cancer IHC. melanoma: 1/11 (9.1%); breast cancer: 0/10 (0.0%)

Biology. May mediate a process in spermatogenesis or may play a role in sex ratio distortion

Curated tissue note. Expressed exclusively in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H322 | HPA portal (live; not the pinned values)

VGLL1 | Unsafe / RNA

ENSG00000102243

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 6.4 nTPM (limit 6); urinary bladder: 21.9 nTPM (limit 12)

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 139/169 (82.2%); Ovary Serous Cystadenocarcinoma (TCGA): 275/349 (78.8%); Ovary Serous Cystadenocarcinoma (validation): 55/81 (67.9%)

Cancer IHC. glioma: 1/12 (8.3%); urothelial cancer: 1/12 (8.3%)

Biology. May act as a specific coactivator for the mammalian TEFs

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q99990 | HPA portal (live; not the pinned values)

VIRMA | Unsafe / Protein

ENSG00000164944

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / basal cells: Medium (Approved); bronchus / ciliated cells (cell body): Medium (Approved); bronchus / ciliated cells (tip of cilia): ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. testis cancer: 11/12 (91.7%); urothelial cancer: 8/11 (72.7%)

Biology. Associated component of the WMM complex, a complex that mediates N6-methyladenosine (m6A) methylation of RNAs, a modification that plays a role in the efficiency of mRNA splicing and RNA processing (PubMed:24981863, PubMed:29507755). Acts as a key regulator of m6A methylation by promoting m6A methylation of mRNAs in the 3'-UTR near the stop codon: recruits the catalytic core components METTL3 and METTL14, thereby guiding m6A ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q69YN4 | HPA portal (live; not the pinned values)

VKORC1 | Unsafe / Protein

ENSG00000167397

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in molecular layer: Low (Approved); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Kidney Renal Clear Cell Carcinoma (TCGA): 521/521 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%)

Cancer IHC. renal cancer: 11/12 (91.7%); liver cancer: 7/12 (58.3%)

Biology. Involved in vitamin K metabolism. Catalytic subunit of the vitamin K epoxide reductase (VKOR) complex which reduces inactive vitamin K 2,3-epoxide to active vitamin K. Vitamin K is required for the gamma-carboxylation of various proteins, including clotting factors, and is required for normal blood coagulation, but also for normal bone development

Curated tissue note. Expressed at highest levels in fetal and adult liver, followed by fetal heart, kidney, and lung, adult heart, and pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BQB6 | HPA portal (live; not the pinned values)

VPREB1 | Unsafe / Protein

ENSG00000169575

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Approved)

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 1/283 (0.4%)

Cancer IHC. testis cancer: 3/10 (30.0%); skin cancer: 1/10 (10.0%)

Biology. Associates with the Ig-mu chain to form a molecular complex that is expressed on the surface of pre-B-cells. This complex presumably regulates Ig gene rearrangements in the early steps of B-cell differentiation

Curated tissue note. Only expressed by pre-B-cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P12018 | HPA portal (live; not the pinned values)

VPS33A | Unsafe / RNA

ENSG00000139719

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 6.6 nTPM (limit 2); amygdala: 5.6 nTPM (limit 2); basal ganglia: 6.4 nTPM (limit 2); bone marrow: 6.3 nTPM (limit 2); cerebellum: 7.5 nTPM (limit 2); cerebral cortex: 7.8 nTPM (limit 2); choroid plexus: 5.7 nTPM (limit 2); colon: 6.6 nTPM (limit 6); duodenum: 5 nTPM (limit 2); heart ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays a role in vesicle-mediated protein trafficking to lysosomal compartments including the endocytic membrane transport and autophagic pathways. Believed to act as a core component of the putative HOPS and CORVET endosomal tethering complexes which are proposed to be involved in the Rab5-to-Rab7 endosome conversion probably implicating MON1A/B, and via binding SNAREs and SNARE complexes to mediate tethering and docking events during ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96AX1 | HPA portal (live; not the pinned values)

VSIG1 | Unsafe / Protein

ENSG00000101842

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. duodenum / glandular cells: Medium (Enhanced); stomach 1 / glandular cells: High (Enhanced); stomach 2 / glandular cells: High (Enhanced); urinary bladder / urothelial cells: High (Enhanced)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 59/80 (73.8%); Pancreatic Adenocarcinoma (TCGA): 110/176 (62.5%); Stomach Adenocarcinoma (TCGA): 193/346 (55.8%)

Cancer IHC. stomach cancer: 6/12 (50.0%); pancreatic cancer: 3/10 (30.0%)

Biology. Detected only in stomach mucosa and testis, and to a much lesser level in pancreas (at protein level). Detected in gastric cancers (31%), esophageal carcinomas (50%) and ovarian cancers (23%)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86XK7 | HPA portal (live; not the pinned values)

VWA3B | Unsafe / Protein

ENSG00000168658

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (cell body): Low (Approved); bronchus / ciliated cells (ciliary rootlets): Medium (Approved); nasopharynx / ciliated cells (cell body): Low (Approved); nasopharynx / ciliated cells (ciliary rootlets): Medium (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 6/58 (10.3%); Lung Adenocarcinoma (TCGA): 26/497 (5.2%); Uterine Corpus Endometrial Carcinoma (TCGA): 8/176 (4.5%)

Cancer IHC. breast cancer: 4/12 (33.3%); skin cancer: 2/11 (18.2%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q502W6 | HPA portal (live; not the pinned values)

WBP2NL | Unsafe / High RNA

ENSG00000183066

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 48 nTPM; unsited/Astrocytes; hpa_single_cell; unresolved; 5.459 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Pancreatic Adenocarcinoma (validation): 8/80 (10.0%); Breast Invasive Carcinoma (validation): 3/50 (6.0%); Glioblastoma Multiforme (validation): 3/58 (5.2%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May play a role in meiotic resumption and pronuclear formation, mediated by a WW domain-signaling pathway during fertilization

Curated tissue note. Expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ICG8 | HPA portal (live; not the pinned values)

WDHD1 | Unsafe / Protein

ENSG00000198554

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / basal cells: Low (Enhanced); colon / glandular cells: Medium (Enhanced); duodenum / glandular cells: High (Enhanced); esophagus / squamous epithelial cells: Medium (Enhanced); kidney / cells in glomeruli: Low (Enhanced); lung / alveolar cells: Low (Enhanced); lymph node / germinal center ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 472/486 (97.1%); Lung Squamous Cell Carcinoma (validation): 65/68 (95.6%)

Cancer IHC. lymphoma: 12/12 (100.0%); testis cancer: 12/12 (100.0%)

Biology. Core replisome component that acts as a replication initiation factor. Binds directly to the CMG complex and functions as a hub to recruit additional proteins to the replication fork

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75717 | HPA portal (live; not the pinned values)

WDPCP | Unsafe / Protein

ENSG00000143951

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: High (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); cerebellum / Purkinje cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. breast cancer: 6/10 (60.0%); pancreatic cancer: 5/12 (41.7%)

Biology. Probable effector of the planar cell polarity signaling pathway which regulates the septin cytoskeleton in both ciliogenesis and collective cell movements. Together with FUZ and WDPCP proposed to function as core component of the CPLANE (ciliogenesis and planar polarity effectors) complex involved in the recruitment of peripheral IFT-A proteins to basal bodies (By similarity). Binds phosphatidylinositol 3-phosphate with highest ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95876 | HPA portal (live; not the pinned values)

WDR35 | Unsafe / Protein

ENSG00000118965

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebral cortex / neuronal cells: Medium (Approved); colon / glandular cells: Medium (Approved); duodenum / glandular cells: High ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 95/100 (95.0%); Pancreatic Adenocarcinoma (validation): 75/80 (93.8%)

Cancer IHC. pancreatic cancer: 6/11 (54.5%); liver cancer: 4/11 (36.4%)

Biology. As a component of the IFT complex A (IFT-A), a complex required for retrograde ciliary transport and entry into cilia of G protein-coupled receptors (GPCRs), it is involved in ciliogenesis and ciliary protein trafficking (PubMed:21473986, PubMed:28400947, PubMed:29220510, PubMed:36775821). May promote CASP3 activation and TNF-stimulated apoptosis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P2L0 | HPA portal (live; not the pinned values)

WDR49 | Unsafe / RNA

ENSG00000174776

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 9.1 nTPM (limit 2); hypothalamus: 2.3 nTPM (limit 2); midbrain: 2.2 nTPM (limit 2); small intestine: 4.3 nTPM (limit 2); spinal cord: 2.3 nTPM (limit 2); choroid plexus: 9.1 nTPM (baseline 2); hypothalamus: 2.3 nTPM (baseline 2); midbrain: 2.2 nTPM (baseline 2); small intestine: 4.3 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Squamous Cell Carcinoma (validation): 10/68 (14.7%); Glioblastoma Multiforme (TCGA): 19/141 (13.5%)

Cancer IHC. liver cancer: 6/12 (50.0%); melanoma: 2/11 (18.2%)

Biology. Associates with components of the nexin-dynein regulatory complex (N-DRC), a key regulator of ciliary/flagellar motility, and might act as an inner dynein arm (IDA) hub or linkage

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IV35 | HPA portal (live; not the pinned values)

WDR53 | Unsafe / Protein

ENSG00000185798

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); bronchus / ciliated cells (ciliary rootlets): Medium (Approved); bronchus / ciliated cells (tip of cilia): Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebral cortex / neuronal cells: Low ... [full text in table]

Cancer RNA. Head and Neck Squamous Cell Carcinoma (TCGA): 492/492 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%)

Cancer IHC. renal cancer: 11/12 (91.7%); testis cancer: 7/11 (63.6%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z5U6 | HPA portal (live; not the pinned values)

WDR62 | Unsafe / RNA

ENSG00000075702

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 5.3 nTPM (limit 2); heart muscle: 21.4 nTPM (limit 12); retina: 6.5 nTPM (limit 2); skeletal muscle: 106.5 nTPM (limit 12); tongue: 60.6 nTPM (limit 6)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 115/133 (86.5%); Breast Invasive Carcinoma (validation): 41/50 (82.0%); Colon Adenocarcinoma (validation): 376/486 (77.4%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. Required for cerebral cortical development. Plays a role in neuronal proliferation and migration (PubMed:20729831, PubMed:20890278). Plays a role in mother-centriole-dependent centriole duplication; the function also seems to involve CEP152, CDK5RAP2 and CEP63 through a stepwise assembled complex at the centrosome that recruits CDK2 required for centriole duplication (PubMed:26297806)

Curated tissue note. Present in fetal brain, enriched within the ventricular and subventricular zone (at protein level). In the embryonic brain it is expressed in mitotic neural precursor cells

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43379 | HPA portal (live; not the pinned values)

WDR64 | Unsafe / High RNA

ENSG00000162843

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Boundary. All audited maxima are within 10% above 30; other gates still require review

Normal RNA / IHC. 30.5 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 2.128 TPM; blood/THSTAR; dice_thstar; vital

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 1/169 (0.6%); Rectum Adenocarcinoma (validation): 1/207 (0.5%); Breast Invasive Carcinoma (TCGA): 1/1022 (0.1%)

Cancer IHC. ovarian cancer: 5/12 (41.7%); breast cancer: 4/12 (33.3%)

Biology. Expressed in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt B1ANS9 | HPA portal (live; not the pinned values)

WDR89 | Unsafe / Protein

ENSG00000140006

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); cerebral cortex / neuronal cells: Low (Uncertain); colon / glandular cells: Medium (Uncertain); colon / peripheral nerve/ganglion: Low (Uncertain); duodenum / glandular cells: Medium ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 484/486 (99.6%)

Cancer IHC. colorectal cancer: 11/12 (91.7%); pancreatic cancer: 10/12 (83.3%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96FK6 | HPA portal (live; not the pinned values)

WDR93 | Unsafe / RNA

ENSG00000140527

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 6.8 nTPM (limit 2); retina: 2.7 nTPM (limit 2); choroid plexus: 6.8 nTPM (baseline 2); retina: 2.7 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 26/50 (52.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 20/176 (11.4%); Glioblastoma Multiforme (validation): 4/58 (6.9%)

Cancer IHC. liver cancer: 4/11 (36.4%); colorectal cancer: 3/12 (25.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6P2C0 | HPA portal (live; not the pinned values)

WFDC11 | Unsafe / RNA

ENSG00000180083

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.9 nTPM (limit 2); bone marrow: 2.9 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 89/207 (43.0%); Colon Adenocarcinoma (validation): 181/486 (37.2%); Stomach Adenocarcinoma (TCGA): 13/346 (3.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEX6 | HPA portal (live; not the pinned values)

WFDC3 | Unsafe / RNA

ENSG00000124116

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. skin: 16.2 nTPM (limit 10); skin: 16.2 nTPM (baseline 10)

Cancer RNA. Breast Invasive Carcinoma (validation): 21/50 (42.0%); Pancreatic Adenocarcinoma (validation): 30/80 (37.5%); Ovary Serous Cystadenocarcinoma (validation): 14/81 (17.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IUB2 | HPA portal (live; not the pinned values)

WFIKKN2 | Unsafe / RNA

ENSG00000173714

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 1135.6 nTPM (limit 2); pituitary gland: 2.2 nTPM (limit 2); skeletal muscle: 2.1 nTPM (limit 2); choroid plexus: 1135.6 nTPM (baseline 2); pituitary gland: 2.2 nTPM (baseline 2); skeletal muscle: 2.1 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 11/133 (8.3%); Glioblastoma Multiforme (validation): 4/58 (6.9%); Glioblastoma Multiforme (TCGA): 6/141 (4.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Protease-inhibitor that contains multiple distinct protease inhibitor domains. Probably has serine protease- and metalloprotease-inhibitor activity. Inhibits the biological activity of mature myostatin, but not activin (By similarity)

Curated tissue note. Primarily expressed in ovary, testis and brain, but not in liver. In fetal tissues, it is primarily expressed in brain, skeletal muscle, thymus and kidney

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TEU8 | HPA portal (live; not the pinned values)

WNK3 | Unsafe / RNA

ENSG00000196632

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pituitary gland: 2.4 nTPM (limit 2); retina: 3.2 nTPM (limit 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Testicular Germ Cell Tumor (TCGA): 24/133 (18.0%); Liver Hepatocellular Carcinoma (validation): 41/231 (17.7%)

Cancer IHC. cervical cancer: 1/11 (9.1%); ovarian cancer: 1/12 (8.3%)

Biology. Serine/threonine-protein kinase component of the WNK3-SPAK/OSR1 kinase cascade, which plays an important role in the regulation of electrolyte homeostasis and regulatory volume increase in response to hyperosmotic stress (PubMed:16275911, PubMed:16275913, PubMed:16501604, PubMed:22989884, PubMed:36318922). WNK3 mediates regulatory volume increase in response to hyperosmotic stress by acting as a molecular crowding sensor, which senses ... [full text in table]

Curated tissue note. Expressed in brain, lung, kidney, liver and pancreas, and in fetal tissues including placenta, fetal brain, lung and kidney. Very low levels of expression were also detected in fetal heart, thymus, liver and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BYP7 | HPA portal (live; not the pinned values)

WNT7A | Unsafe / RNA

ENSG00000154764

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.7 nTPM (limit 2); basal ganglia: 3.2 nTPM (limit 2); amygdala: 3.7 nTPM (baseline 2); basal ganglia: 3.2 nTPM (baseline 2); cerebral cortex: 4 nTPM (baseline 2)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 229/349 (65.6%); Ovary Serous Cystadenocarcinoma (validation): 51/81 (63.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 92/176 (52.3%)

Cancer IHC. prostate cancer: 4/12 (33.3%); glioma: 2/12 (16.7%)

Biology. Ligand for members of the frizzled family of seven transmembrane receptors that functions in the canonical Wnt/beta-catenin signaling pathway (By similarity). Plays an important role in embryonic development, including dorsal versus ventral patterning during limb development, skeleton development and urogenital tract development (PubMed:16826533). Required for central nervous system (CNS) angiogenesis and blood-brain barrier regulation ... [full text in table]

Curated tissue note. Expression is restricted to placenta, kidney, testis, uterus, fetal lung, and fetal and adult brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00755 | HPA portal (live; not the pinned values)

XAGE2 | Unsafe / RNA

ENSG00000155622

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. small intestine / glandular cells: Low (Supported); stomach 1 / glandular cells: Low (Supported); thyroid gland / glandular cells: Low (Supported)

Cancer RNA. Ovary Serous Cystadenocarcinoma (TCGA): 93/349 (26.6%); Lung Adenocarcinoma (validation): 23/105 (21.9%); Lung Adenocarcinoma (TCGA): 80/497 (16.1%)

Cancer IHC. urothelial cancer: 2/12 (16.7%); prostate cancer: 1/10 (10.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96GT9 | HPA portal (live; not the pinned values)

XAGE3 | Unsafe / High RNA

ENSG00000171402

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 68.6 nTPM; placenta/t-cells; hpa_single_cell_tissue; vital; 0.8567 TPM; blood/memory B-cell; hpa_immune_cell_samples; vital

Cancer RNA. Skin Cuteneous Melanoma (TCGA): 6/99 (6.1%); Ovary Serous Cystadenocarcinoma (TCGA): 14/349 (4.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 5/176 (2.8%)

Cancer IHC. urothelial cancer: 2/12 (16.7%); prostate cancer: 1/10 (10.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WTP9 | HPA portal (live; not the pinned values)

XKR9 | Unsafe / Protein

ENSG00000221947

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); cerebral cortex / neuronal cells: Medium (Enhanced); colon / glandular cells: Low (Enhanced); ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 72/80 (90.0%); Lung Adenocarcinoma (validation): 30/105 (28.6%); Colon Adenocarcinoma (validation): 131/486 (27.0%)

Cancer IHC. prostate cancer: 4/10 (40.0%); endometrial cancer: 3/10 (30.0%)

Biology. Phospholipid scramblase that promotes phosphatidylserine exposure on apoptotic cell surface (PubMed:25231987). Phosphatidylserine is a specific marker only present at the surface of apoptotic cells and acts as a specific signal for engulfment (PubMed:25231987)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5GH70 | HPA portal (live; not the pinned values)

XPO4 | Unsafe / RNA

ENSG00000132953

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.5 nTPM (limit 2); bone marrow: 10.1 nTPM (limit 2); cerebellum: 4.7 nTPM (limit 2); cerebral cortex: 4 nTPM (limit 2); choroid plexus: 2.8 nTPM (limit 2); duodenum: 3.6 nTPM (limit 2); heart muscle: 10.4 nTPM (limit 2); kidney: 4.9 nTPM (limit 2); liver: 4 nTPM (limit 2); lung: 5.9 ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 484/486 (99.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Mediates the nuclear export of proteins (cargos), such as EIF5A, SMAD3 and isoform M2 of PKM (PKM2) (PubMed:10944119, PubMed:16449645, PubMed:26787900). In the nucleus binds cooperatively to its cargo and to the GTPase Ran in its active GTP-bound form. Docking of this trimeric complex to the nuclear pore complex (NPC) is mediated through binding to nucleoporins (PubMed:10944119, PubMed:16449645). Upon transit of a nuclear export ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9C0E2 | HPA portal (live; not the pinned values)

XRCC2 | Unsafe / RNA

ENSG00000196584

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 5.7 nTPM (limit 2); bone marrow: 5.7 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Colon Adenocarcinoma (validation): 460/486 (94.7%); Rectum Adenocarcinoma (validation): 192/207 (92.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Involved in the homologous recombination repair (HRR) pathway of double-stranded DNA, thought to repair chromosomal fragmentation, translocations and deletions. Part of the RAD51 paralog protein complex BCDX2 which acts in the BRCA1-BRCA2-dependent HR pathway. Upon DNA damage, BCDX2 acts downstream of BRCA2 recruitment and upstream of RAD51 recruitment. BCDX2 binds predominantly to the intersection of the four duplex arms of the ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43543 | HPA portal (live; not the pinned values)

XRCC4 | Unsafe / Protein

ENSG00000152422

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Supported); adrenal gland / glandular cells: High (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / glial cells: High (Supported); caudate / neuronal cells: Medium (Supported); ... [full text in table]

Cancer RNA. Lung Adenocarcinoma (validation): 105/105 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. lymphoma: 12/12 (100.0%); breast cancer: 11/11 (100.0%)

Biology. DNA non-homologous end joining (NHEJ) core factor, required for double-strand break repair and V(D)J recombination (PubMed:10757784, PubMed:10854421, PubMed:12517771, PubMed:16412978, PubMed:17124166, PubMed:17290226, PubMed:22228831, PubMed:25597996, PubMed:25742519, PubMed:25934149, PubMed:26100018, PubMed:26774286, PubMed:8548796). Acts as a scaffold protein that regulates recruitment of other proteins to DNA double-strand breaks ... [full text in table]

Curated tissue note. Widely expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q13426 | HPA portal (live; not the pinned values)

YEATS4 | Unsafe / RNA

ENSG00000127337

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 11.5 nTPM (limit 2); amygdala: 8.7 nTPM (limit 2); basal ganglia: 8.8 nTPM (limit 2); bone marrow: 45.2 nTPM (limit 2); cerebellum: 16.6 nTPM (limit 2); cerebral cortex: 10.6 nTPM (limit 2); choroid plexus: 14.9 nTPM (limit 2); colon: 11.9 nTPM (limit 6); duodenum: 13.1 nTPM (limit ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Ovary Serous Cystadenocarcinoma (TCGA): 349/349 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Chromatin reader component of the NuA4 histone acetyltransferase (HAT) complex, a complex involved in transcriptional activation of select genes principally by acetylation of nucleosomal histones H4 and H2A (PubMed:12963728, PubMed:14966270). Specifically recognizes and binds acylated histone H3, with a preference for histone H3 diacetylated at 'Lys-18' and 'Lys-27' (H3K18ac and H3K27ac) or histone H3 diacetylated at 'Lys-14' and ... [full text in table]

Curated tissue note. Expressed in brain, heart, kidney, liver, lung, pancreas, placenta and skeletal muscle

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95619 | HPA portal (live; not the pinned values)

YPEL1 | Unsafe / Protein

ENSG00000100027

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Prostate Adenocarcinoma (TCGA): 457/480 (95.2%); Breast Invasive Carcinoma (validation): 45/50 (90.0%)

Cancer IHC. liver cancer: 12/12 (100.0%); colorectal cancer: 11/12 (91.7%)

Biology. May play a role in epithelioid conversion of fibroblasts

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O60688 | HPA portal (live; not the pinned values)

ZBBX | Unsafe / Protein

ENSG00000169064

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / ciliated cells (ciliary rootlets): Low (Enhanced); bronchus / ciliated cells (tip of cilia): High (Enhanced); nasopharynx / ciliated cells (cilia axoneme): Medium (Enhanced); nasopharynx / ciliated cells (tip of cilia): High (Enhanced)

Cancer RNA. Glioblastoma Multiforme (validation): 21/58 (36.2%); Ovary Serous Cystadenocarcinoma (validation): 14/81 (17.3%); Breast Invasive Carcinoma (validation): 5/50 (10.0%)

Cancer IHC. breast cancer: 0/9 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A8MT70 | HPA portal (live; not the pinned values)

ZBTB20 | Unsafe / Protein

ENSG00000181722

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Approved); adrenal gland / glandular cells: High (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low (Approved); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. glioma: 11/12 (91.7%); renal cancer: 8/12 (66.7%)

Biology. May be a transcription factor that may be involved in hematopoiesis, oncogenesis, and immune responses (PubMed:11352661). Plays a role in postnatal myogenesis, may be involved in the regulation of satellite cells self-renewal (By similarity)

Curated tissue note. Expressed in spleen, lymph node, thymus, peripheral blood leukocytes, and fetal liver

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HC78 | HPA portal (live; not the pinned values)

ZBTB3 | Unsafe / Protein

ENSG00000185670

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 196/207 (94.7%); Colon Adenocarcinoma (validation): 455/486 (93.6%)

Cancer IHC. liver cancer: 11/11 (100.0%); ovarian cancer: 11/11 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H5J0 | HPA portal (live; not the pinned values)

ZBTB32 | Unsafe / Protein

ENSG00000011590

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); lung / macrophages: Medium (Enhanced); lymph node / non-germinal center cells: Low (Enhanced); skin 1 / Langerhans: Medium (Enhanced); spleen / cells in red pulp: Low (Enhanced)

Cancer RNA. Breast Invasive Carcinoma (validation): 8/50 (16.0%); Pancreatic Adenocarcinoma (validation): 2/80 (2.5%); Testicular Germ Cell Tumor (TCGA): 3/133 (2.3%)

Cancer IHC. lymphoma: 3/11 (27.3%); breast cancer: 0/11 (0.0%)

Biology. DNA-binding protein that binds to the to a 5'-TGTACAGTGT-3' core sequence. May function as a transcriptional transactivator and transcriptional repressor. Probably exerts its repressor effect by preventing GATA3 from binding to DNA. May play a role in regulating the differentiation and activation of helper T-cells (By similarity)

Curated tissue note. Predominantly expressed in testis. Some isoforms are ubiquitously expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y2Y4 | HPA portal (live; not the pinned values)

ZBTB37 | Unsafe / Protein

ENSG00000185278

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 447/486 (92.0%); Rectum Adenocarcinoma (validation): 185/207 (89.4%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); prostate cancer: 11/11 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5TC79 | HPA portal (live; not the pinned values)

ZBTB8A | Unsafe / Protein

ENSG00000160062

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 45/50 (90.0%); Pancreatic Adenocarcinoma (validation): 71/80 (88.8%); Glioblastoma Multiforme (validation): 45/58 (77.6%)

Cancer IHC. pancreatic cancer: 9/10 (90.0%); stomach cancer: 7/8 (87.5%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96BR9 | HPA portal (live; not the pinned values)

ZBTB9 | Unsafe / RNA

ENSG00000213588

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.8 nTPM (limit 2); basal ganglia: 2.4 nTPM (limit 2); cerebellum: 2.7 nTPM (limit 2); cerebral cortex: 2.7 nTPM (limit 2); choroid plexus: 2.6 nTPM (limit 2); duodenum: 2.3 nTPM (limit 2); heart muscle: 2.9 nTPM (limit 2); hypothalamus: 2.6 nTPM (limit 2); kidney: 4.3 nTPM (limit ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 48/50 (96.0%); Ovary Serous Cystadenocarcinoma (validation): 61/81 (75.3%)

Cancer IHC. liver cancer: 10/12 (83.3%); prostate cancer: 10/12 (83.3%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96C00 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ZC2HC1C | Unsafe / Protein

ENSG00000119703

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); cerebral cortex / neuronal cells: Low (Enhanced); cerebral cortex / neuropil: Medium (Enhanced); hippocampus / neuronal cells: Low (Enhanced); smooth muscle / smooth muscle cells: Low (Enhanced)

Cancer RNA. Kidney Chromophobe (TCGA): 49/64 (76.6%); Thyroid Carcinoma (TCGA): 372/495 (75.2%); Kidney Renal Papillary Cell Carcinoma (TCGA): 162/282 (57.4%)

Cancer IHC. breast cancer: 0/10 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q53FD0 | HPA portal (live; not the pinned values)

ZCWPW1 | Unsafe / RNA

ENSG00000078487

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.5 nTPM (limit 2); amygdala: 3.5 nTPM (limit 2); basal ganglia: 3.1 nTPM (limit 2); bone marrow: 2.8 nTPM (limit 2); cerebral cortex: 3.2 nTPM (limit 2); choroid plexus: 4 nTPM (limit 2); heart muscle: 3.7 nTPM (limit 2); hippocampal formation: 3.6 nTPM (limit 2); hypothalamus: 3 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Ovary Serous Cystadenocarcinoma (validation): 38/81 (46.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Dual histone methylation reader specific for PRDM9-catalyzed histone marks (H3K4me3 and H3K36me3) (PubMed:20826339, PubMed:32744506). Facilitates the repair of PRDM9-induced meiotic double-strand breaks (DSBs) (By similarity). Essential for male fertility and spermatogenesis (By similarity). Required for meiosis prophase I progression in male but not in female germ cells (By similarity)

Curated tissue note. Testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0M4 | HPA portal (live; not the pinned values)

ZDHHC15 | Unsafe / Protein

ENSG00000102383

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 75/141 (53.2%); Breast Invasive Carcinoma (validation): 22/50 (44.0%)

Cancer IHC. liver cancer: 8/11 (72.7%); urothelial cancer: 8/12 (66.7%)

Biology. Palmitoyltransferase that catalyzes the addition of palmitate onto various protein substrates (PubMed:18817523, PubMed:23034182). Has no stringent fatty acid selectivity and in addition to palmitate can also transfer onto target proteins myristate from tetradecanoyl-CoA and stearate from octadecanoyl-CoA (By similarity). Palmitoylates IGF2R and SORT1, promoting their partitioning to an endosomal membrane subdomain where they can ... [full text in table]

Curated tissue note. Expressed in placenta, liver, lung, kidney, heart and brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96MV8 | HPA portal (live; not the pinned values)

ZDHHC19 | Unsafe / High RNA

ENSG00000163958

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 170 nTPM; salivary gland/serous glandular cells; hpa_single_cell_tissue; protected; 22.36 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Liver Hepatocellular Carcinoma (validation): 24/231 (10.4%); Ovary Serous Cystadenocarcinoma (validation): 1/81 (1.2%); Testicular Germ Cell Tumor (TCGA): 1/133 (0.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Palmitoyltransferase that mediates palmitoylation oproteins, such as RRAS and SQSTM1 (PubMed:20074548, PubMed:37802024). Catalyzes palmitoylation of RRAS, leading to increased cell viability (PubMed:20074548). Acts as a positive regulator of autophagy by mediating palmitoylation of SQSTM1, promoting affinity between SQSTM1 and ATG8 proteins and recruitment of ubiquitinated cargo proteins to autophagosomes (PubMed:37802024). ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8WVZ1 | HPA portal (live; not the pinned values)

ZDHHC20 | Unsafe / Protein

ENSG00000180776

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); endometrial cancer: 11/11 (100.0%)

Biology. Palmitoyltransferase that could catalyze the addition of palmitate onto various protein substrates (PubMed:27153536, PubMed:29326245, PubMed:33219126). Catalyzes palmitoylation of Cys residues in the cytoplasmic C-terminus of EGFR, and modulates the duration of EGFR signaling by modulating palmitoylation-dependent EGFR internalization and degradation (PubMed:27153536). Has a preference for acyl-CoA with C16 fatty acid chains ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5W0Z9 | HPA portal (live; not the pinned values)

ZFAND4 | Unsafe / Protein

ENSG00000172671

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. duodenum / glandular cells: Low (Enhanced); kidney / cells in tubules: Low (Enhanced); small intestine / glandular cells: Low (Enhanced); stomach 1 / glandular cells: Low (Enhanced)

Cancer RNA. Pancreatic Adenocarcinoma (validation): 75/80 (93.8%); Glioblastoma Multiforme (validation): 52/58 (89.7%); Ovary Serous Cystadenocarcinoma (validation): 29/81 (35.8%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86XD8 | HPA portal (live; not the pinned values)

ZFP37 | Unsafe / RNA

ENSG00000136866

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.1 nTPM (limit 2); cerebellum: 4.2 nTPM (limit 2); cerebral cortex: 3.5 nTPM (limit 2); hypothalamus: 2.1 nTPM (limit 2); kidney: 2.2 nTPM (limit 2); pituitary gland: 3.7 nTPM (limit 2); retina: 6.1 nTPM (limit 2); smooth muscle: 2.4 nTPM (limit 2); spinal cord: 2.4 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 37/50 (74.0%); Glioblastoma Multiforme (TCGA): 72/141 (51.1%)

Cancer IHC. breast cancer: 0/11 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May be involved in transcriptional regulation

Curated tissue note. Expressed at low level in several tissues including fetal cartilage

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y6Q3 | HPA portal (live; not the pinned values)

ZFP64 | Unsafe / RNA

ENSG00000020256

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.7 nTPM (limit 2); amygdala: 3.9 nTPM (limit 2); basal ganglia: 4.3 nTPM (limit 2); bone marrow: 5 nTPM (limit 2); cerebellum: 4.6 nTPM (limit 2); cerebral cortex: 6 nTPM (limit 2); choroid plexus: 3.4 nTPM (limit 2); duodenum: 6.3 nTPM (limit 2); esophagus: 6.8 nTPM (limit 6); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Rectum Adenocarcinoma (TCGA): 88/88 (100.0%)

Cancer IHC. urothelial cancer: 1/11 (9.1%); ovarian cancer: 1/12 (8.3%)

Biology. May be involved in the regulation of mesenchymal cell differentiation through transactivation of NOTCH1 target genes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NTW7 | HPA portal (live; not the pinned values)

ZFP69 | Unsafe / RNA

ENSG00000187815

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 3.1 nTPM (limit 2); basal ganglia: 3.2 nTPM (limit 2); cerebellum: 2.3 nTPM (limit 2); cerebral cortex: 2.3 nTPM (limit 2); hippocampal formation: 2.9 nTPM (limit 2); hypothalamus: 2.5 nTPM (limit 2); kidney: 2.1 nTPM (limit 2); lung: 2.3 nTPM (limit 2); midbrain: 3 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 49/58 (84.5%); Testicular Germ Cell Tumor (TCGA): 67/133 (50.4%); Glioblastoma Multiforme (TCGA): 49/141 (34.8%)

Cancer IHC. liver cancer: 4/12 (33.3%); stomach cancer: 3/11 (27.3%)

Biology. Putative transcription factor that appears to regulate lipid metabolism

Curated tissue note. Expressed in visceral and subcutaneous adipose tissue

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q49AA0 | HPA portal (live; not the pinned values)

ZFPL1 | Unsafe / Protein

ENSG00000162300

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Enhanced); adrenal gland / glandular cells: Medium (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. endometrial cancer: 11/12 (91.7%); glioma: 11/12 (91.7%)

Biology. Required for cis-Golgi integrity and efficient ER to Golgi transport. Involved in the maintenance of the integrity of the cis-Golgi, possibly via its interaction with GOLGA2/GM130

Curated tissue note. Expressed strongly in the exocrine pancreas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95159 | HPA portal (live; not the pinned values)

ZFX | Unsafe / Protein

ENSG00000005889

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Supported); adrenal gland / glandular cells: Low (Supported); bone marrow / hematopoietic cells: Medium (Supported); bronchus / respiratory epithelial cells: Low (Supported); caudate / glial cells: Low (Supported); caudate / neuronal cells: Low (Supported); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. breast cancer: 9/12 (75.0%); prostate cancer: 6/12 (50.0%)

Biology. Probable transcriptional activator

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P17010 | HPA portal (live; not the pinned values)

ZFY | Unsafe / Protein

ENSG00000067646

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / cells in molecular layer: Medium (Uncertain); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 33/58 (56.9%); Rectum Adenocarcinoma (validation): 108/207 (52.2%); Colon Adenocarcinoma (validation): 221/486 (45.5%)

Cancer IHC. breast cancer: 9/12 (75.0%); prostate cancer: 6/12 (50.0%)

Biology. Probable transcriptional activator. Binds to the consensus sequence 5'-AGGCCY-3'

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P08048 | HPA portal (live; not the pinned values)

ZIC2 | Unsafe / RNA

ENSG00000043355

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 5.9 nTPM (limit 2); basal ganglia: 4 nTPM (limit 2); cerebellum: 155.1 nTPM (limit 2); cerebral cortex: 9.9 nTPM (limit 2); choroid plexus: 19.5 nTPM (limit 2); hippocampal formation: 7.9 nTPM (limit 2); hypothalamus: 5.6 nTPM (limit 2); midbrain: 3.7 nTPM (limit 2); retina: 2.4 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 32/58 (55.2%); Glioblastoma Multiforme (TCGA): 70/141 (49.6%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 139/283 (49.1%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Acts as a transcriptional activator or repressor. Plays important roles in the early stage of organogenesis of the CNS. Activates the transcription of the serotonin transporter SERT in uncrossed ipsilateral retinal ganglion cells (iRGCs) to refine eye-specific projections in primary visual targets. Its transcriptional activity is repressed by MDFIC. Involved in the formation of the ipsilateral retinal projection at the optic chiasm ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95409 | HPA portal (live; not the pinned values)

ZIC5 | Unsafe / RNA

ENSG00000139800

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 27 nTPM (limit 2); cerebral cortex: 2.3 nTPM (limit 2); choroid plexus: 5.3 nTPM (limit 2); retina: 3.3 nTPM (limit 2); cerebellum: 27 nTPM (baseline 2); cerebral cortex: 2.3 nTPM (baseline 2); choroid plexus: 5.3 nTPM (baseline 2); retina: 3.3 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 32/58 (55.2%); Colon Adenocarcinoma (validation): 119/486 (24.5%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 43/283 (15.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Essential for neural crest development, converting cells from an epidermal fate to a neural crest cell fate. Binds to DNA (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96T25 | HPA portal (live; not the pinned values)

ZKSCAN2 | Unsafe / RNA

ENSG00000155592

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.2 nTPM (limit 2); retina: 4 nTPM (limit 2); skeletal muscle: 3.1 nTPM (limit 2); cerebral cortex: 2.2 nTPM (baseline 2); retina: 4 nTPM (baseline 2); skeletal muscle: 3.1 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 49/50 (98.0%); Testicular Germ Cell Tumor (TCGA): 38/133 (28.6%)

Cancer IHC. cervical cancer: 4/11 (36.4%); colorectal cancer: 4/12 (33.3%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q63HK3 | HPA portal (live; not the pinned values)

ZKSCAN3 | Unsafe / Protein

ENSG00000189298

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: Low (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Rectum Adenocarcinoma (validation): 153/207 (73.9%)

Cancer IHC. cervical cancer: 11/12 (91.7%); lymphoma: 11/12 (91.7%)

Biology. Transcriptional factor that binds to the consensus sequence 5'-[GT][AG][AGT]GGGG-3' and acts as a repressor of autophagy. Specifically represses expression of genes involved in autophagy and lysosome biogenesis/function such as MAP1LC3B, ULK1 or WIPI2. Associates with chromatin at the ITGB4 and VEGF promoters. Also acts as a transcription activator and promotes cancer cell progression and/or migration in various tumors and myelomas

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BRR0 | HPA portal (live; not the pinned values)

ZMYM5 | Unsafe / Protein

ENSG00000132950

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / Purkinje cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. renal cancer: 12/12 (100.0%); pancreatic cancer: 11/11 (100.0%)

Biology. Functions as a transcriptional regulator

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UJ78 | HPA portal (live; not the pinned values)

ZMYM6 | Unsafe / Protein

ENSG00000163867

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: High (Approved); ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Rectum Adenocarcinoma (validation): 206/207 (99.5%); Colon Adenocarcinoma (validation): 482/486 (99.2%)

Cancer IHC. glioma: 12/12 (100.0%); lymphoma: 11/12 (91.7%)

Biology. Required for regulation of cell morphology and cytoskeletal organization

Curated tissue note. Expressed at high levels in heart, skeletal muscle, kidney and liver

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O95789 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ZMYND10 | Unsafe / RNA

ENSG00000004838

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 5.8 nTPM (limit 2); basal ganglia: 12.3 nTPM (limit 2); choroid plexus: 82.5 nTPM (limit 2); hippocampal formation: 8.2 nTPM (limit 2); hypothalamus: 10.8 nTPM (limit 2); kidney: 2.1 nTPM (limit 2); lung: 10.8 nTPM (limit 2); pituitary gland: 5.8 nTPM (limit 2); retina: 3.1 nTPM (limit ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 48/50 (96.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 198/282 (70.2%); Breast Invasive Carcinoma (TCGA): 636/1022 (62.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Plays a role in axonemal structure organization and motility (PubMed:23891469, PubMed:23891471). Involved in axonemal pre-assembly of inner and outer dynein arms (IDA and ODA, respectively) for proper axoneme building for cilia motility (By similarity). May act by indirectly regulating transcription of dynein proteins (By similarity)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O75800 | HPA portal (live; not the pinned values)

ZMYND12 | Unsafe / RNA

ENSG00000066185

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.2 nTPM (limit 2); amygdala: 3 nTPM (limit 2); basal ganglia: 4 nTPM (limit 2); cerebral cortex: 2.5 nTPM (limit 2); choroid plexus: 9.9 nTPM (limit 2); heart muscle: 2.2 nTPM (limit 2); hippocampal formation: 2.8 nTPM (limit 2); hypothalamus: 4.3 nTPM (limit 2); kidney: 9 nTPM ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 36/50 (72.0%); Glioblastoma Multiforme (validation): 24/58 (41.4%); Ovary Serous Cystadenocarcinoma (TCGA): 143/349 (41.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Required for sperm flagellum function and male fertility

Curated tissue note. Expressed predominantly in the testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0C1 | HPA portal (live; not the pinned values)

ZMYND15 | Unsafe / RNA

ENSG00000141497

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 2.3 nTPM (limit 2); duodenum: 3.9 nTPM (limit 2); liver: 3 nTPM (limit 2); lung: 4.5 nTPM (limit 2); small intestine: 2.9 nTPM (limit 2); spinal cord: 3.2 nTPM (limit 2); choroid plexus: 2.3 nTPM (baseline 2); duodenum: 3.9 nTPM (baseline 2); liver: 3 nTPM (baseline 2); lung: 4.5 ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 40/50 (80.0%); Kidney Renal Papillary Cell Carcinoma (TCGA): 102/282 (36.2%); Liver Hepatocellular Carcinoma (validation): 83/231 (35.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Acts as a transcriptional repressor through interaction with histone deacetylases (HDACs). May be important for spermiogenesis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H091 | HPA portal (live; not the pinned values)

ZNF10 | Unsafe / RNA

ENSG00000256223

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 13.8 nTPM (limit 2); amygdala: 4.5 nTPM (limit 2); basal ganglia: 4.9 nTPM (limit 2); bone marrow: 8.5 nTPM (limit 2); cerebellum: 6.7 nTPM (limit 2); cerebral cortex: 5.6 nTPM (limit 2); choroid plexus: 11.7 nTPM (limit 2); colon: 10.6 nTPM (limit 6); duodenum: 3.6 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Thyroid Carcinoma (TCGA): 446/495 (90.1%); Breast Invasive Carcinoma (validation): 45/50 (90.0%)

Cancer IHC. prostate cancer: 9/10 (90.0%); colorectal cancer: 6/10 (60.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P21506 | HPA portal (live; not the pinned values)

ZNF101 | Unsafe / RNA

ENSG00000181896

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.5 nTPM (limit 2); basal ganglia: 2.4 nTPM (limit 2); bone marrow: 13.4 nTPM (limit 2); cerebellum: 2.3 nTPM (limit 2); cerebral cortex: 2.6 nTPM (limit 2); choroid plexus: 3 nTPM (limit 2); duodenum: 3.1 nTPM (limit 2); esophagus: 6.7 nTPM (limit 6); heart muscle: 2.1 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 203/207 (98.1%); Colon Adenocarcinoma (validation): 476/486 (97.9%)

Cancer IHC. melanoma: 1/11 (9.1%); breast cancer: 1/12 (8.3%)

Biology. May be involved in transcriptional regulation

Curated tissue note. Expressed in a variety of adult and fetal tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZC7 | HPA portal (live; not the pinned values)

ZNF112 | Unsafe / Protein

ENSG00000062370

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebral cortex / endothelial cells: Low (Approved); cerebral cortex / neuropil: Low (Approved); colon / endothelial cells: Low (Approved); colon / glandular cells: Low (Approved); colon / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 47/80 (58.8%); Glioblastoma Multiforme (TCGA): 66/141 (46.8%)

Cancer IHC. melanoma: 4/12 (33.3%); liver cancer: 2/12 (16.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UJU3 | HPA portal (live; not the pinned values)

ZNF124 | Unsafe / RNA

ENSG00000196418

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 4.5 nTPM (limit 2); duodenum: 5.3 nTPM (limit 2); kidney: 3.9 nTPM (limit 2); liver: 2.7 nTPM (limit 2); lung: 3.4 nTPM (limit 2); pancreas: 3.3 nTPM (limit 2); retina: 3.1 nTPM (limit 2); small intestine: 5.2 nTPM (limit 2); smooth muscle: 2.9 nTPM (limit 2); bone marrow: 4.5 nTPM ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); endometrial cancer: 9/11 (81.8%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15973 | HPA portal (live; not the pinned values)

ZNF165 | Unsafe / RNA

ENSG00000197279

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. choroid plexus: 4.9 nTPM (limit 2); duodenum: 2.9 nTPM (limit 2); kidney: 4.4 nTPM (limit 2); liver: 2.4 nTPM (limit 2); lung: 2.5 nTPM (limit 2); pancreas: 4.6 nTPM (limit 2); parathyroid gland: 6.2 nTPM (limit 6); pituitary gland: 3.7 nTPM (limit 2); retina: 2.2 nTPM (limit 2); small intestine: ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 47/50 (94.0%); Colon Adenocarcinoma (validation): 410/486 (84.4%); Rectum Adenocarcinoma (validation): 147/207 (71.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Curated tissue note. Expressed specifically in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P49910 | HPA portal (live; not the pinned values)

ZNF169 | Unsafe / Protein

ENSG00000175787

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: High (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 200/207 (96.6%); Colon Adenocarcinoma (validation): 469/486 (96.5%); Breast Invasive Carcinoma (validation): 47/50 (94.0%)

Cancer IHC. lymphoma: 12/12 (100.0%); breast cancer: 11/11 (100.0%)

Biology. May be involved in transcriptional regulation

Curated tissue note. Highly expressed in kidney, weakly expressed in heart, liver, spleen, and small intestine. Not expressed in adult brain or spinal cord

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14929 | HPA portal (live; not the pinned values)

ZNF174 | Unsafe / Protein

ENSG00000103343

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: High (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. glioma: 10/12 (83.3%); melanoma: 9/12 (75.0%)

Biology. Transcriptional repressor

Curated tissue note. Expressed in a variety of organs, but most strongly in adult testis and ovary followed by small intestine, colon, prostate, thymus, spleen, pancreas, skeletal muscle, heart, brain and kidney. Also expressed in ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q15697 | HPA portal (live; not the pinned values)

ZNF19 | Unsafe / Protein

ENSG00000157429

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Approved); caudate / glial cells: Low (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / cells in molecular layer: Low (Approved); colon / endothelial cells: Medium (Approved); colon / glandular cells: Medium (Approved); duodenum ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 203/207 (98.1%); Colon Adenocarcinoma (validation): 469/486 (96.5%); Breast Invasive Carcinoma (validation): 46/50 (92.0%)

Cancer IHC. liver cancer: 6/10 (60.0%); prostate cancer: 7/12 (58.3%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P17023 | HPA portal (live; not the pinned values)

ZNF2 | Unsafe / RNA

ENSG00000275111

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.5 nTPM (limit 2); cerebellum: 2.2 nTPM (limit 2); cerebral cortex: 2.4 nTPM (limit 2); heart muscle: 2.9 nTPM (limit 2); kidney: 2.5 nTPM (limit 2); lung: 2.3 nTPM (limit 2); pancreas: 2.1 nTPM (limit 2); retina: 2.7 nTPM (limit 2); skeletal muscle: 2.9 nTPM (limit 2); smooth ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Testicular Germ Cell Tumor (TCGA): 57/133 (42.9%); Rectum Adenocarcinoma (validation): 80/207 (38.6%)

Cancer IHC. ovarian cancer: 9/12 (75.0%); pancreatic cancer: 7/10 (70.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BSG1 | HPA portal (live; not the pinned values)

ZNF208 | Unsafe / RNA

ENSG00000160321

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.2 nTPM (limit 2); basal ganglia: 2.4 nTPM (limit 2); cerebral cortex: 2.8 nTPM (limit 2); heart muscle: 10.9 nTPM (limit 2); hippocampal formation: 4 nTPM (limit 2); hypothalamus: 8.7 nTPM (limit 2); kidney: 4.4 nTPM (limit 2); midbrain: 8 nTPM (limit 2); pancreas: 2.1 nTPM (limit 2); ... [full text in table]

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 123/133 (92.5%); Thyroid Carcinoma (TCGA): 429/495 (86.7%); Rectum Adenocarcinoma (validation): 117/207 (56.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Curated tissue note. Ubiquitous

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43345 | HPA portal (live; not the pinned values)

ZNF214 | Unsafe / RNA

ENSG00000149050

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 2.3 nTPM (limit 2); retina: 5.2 nTPM (limit 2); kidney: 2.3 nTPM (baseline 2); retina: 5.2 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 46/58 (79.3%); Breast Invasive Carcinoma (validation): 34/50 (68.0%); Pancreatic Adenocarcinoma (validation): 12/80 (15.0%)

Cancer IHC. colorectal cancer: 4/11 (36.4%); breast cancer: 1/11 (9.1%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UL59 | HPA portal (live; not the pinned values)

ZNF215 | Unsafe / Protein

ENSG00000149054

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Enhanced); heart muscle / cardiomyocytes: Low (Enhanced); lung / alveolar cells: Low (Enhanced); lymph node / non-germinal center cells: Low (Enhanced); pancreas / exocrine glandular cells: Low (Enhanced); skin 2 / epidermal cells: Low (Enhanced); smooth ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 139/207 (67.1%); Colon Adenocarcinoma (validation): 313/486 (64.4%); Pancreatic Adenocarcinoma (validation): 20/80 (25.0%)

Cancer IHC. liver cancer: 1/9 (11.1%); breast cancer: 0/12 (0.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UL58 | HPA portal (live; not the pinned values)

ZNF221 | Unsafe / RNA

ENSG00000159905

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 2.5 nTPM (limit 2); retina: 2.9 nTPM (limit 2); cerebellum: 2.5 nTPM (baseline 2); retina: 2.9 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 34/80 (42.5%); Breast Invasive Carcinoma (validation): 13/50 (26.0%)

Cancer IHC. breast cancer: 4/12 (33.3%); ovarian cancer: 2/11 (18.2%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UK13 | HPA portal (live; not the pinned values)

ZNF223 | Unsafe / Protein

ENSG00000178386

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); colon / glandular cells: Medium (Approved); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Glioblastoma Multiforme (validation): 47/58 (81.0%); Rectum Adenocarcinoma (validation): 167/207 (80.7%)

Cancer IHC. stomach cancer: 8/11 (72.7%); urothelial cancer: 6/12 (50.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UK11 | HPA portal (live; not the pinned values)

ZNF225 | Unsafe / RNA

ENSG00000256294

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.1 nTPM (limit 2); bone marrow: 2.3 nTPM (limit 2); cerebellum: 3.2 nTPM (limit 2); cerebral cortex: 3.3 nTPM (limit 2); choroid plexus: 3.8 nTPM (limit 2); duodenum: 2.4 nTPM (limit 2); heart muscle: 3.1 nTPM (limit 2); kidney: 2.9 nTPM (limit 2); liver: 2.7 nTPM (limit 2); lung: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 48/50 (96.0%); Lung Adenocarcinoma (validation): 59/105 (56.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UK10 | HPA portal (live; not the pinned values)

ZNF227 | Unsafe / Protein

ENSG00000131115

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in molecular layer: Medium (Uncertain); cerebellum / Purkinje cells: Medium (Uncertain); cerebral cortex / glial cells: Low (Uncertain); cerebral cortex / neuronal cells: Medium ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. colorectal cancer: 4/12 (33.3%); liver cancer: 2/11 (18.2%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86WZ6 | HPA portal (live; not the pinned values)

ZNF229 | Unsafe / RNA

ENSG00000278318

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 4.3 nTPM (limit 2); retina: 4.3 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 32/50 (64.0%); Testicular Germ Cell Tumor (TCGA): 46/133 (34.6%); Kidney Chromophobe (TCGA): 7/64 (10.9%)

Cancer IHC. glioma: 7/11 (63.6%); melanoma: 5/8 (62.5%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UJW7 | HPA portal (live; not the pinned values)

ZNF23 | Unsafe / RNA

ENSG00000167377

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.1 nTPM (limit 2); amygdala: 5.5 nTPM (limit 2); basal ganglia: 5.6 nTPM (limit 2); bone marrow: 4.3 nTPM (limit 2); cerebellum: 10.5 nTPM (limit 2); cerebral cortex: 6.4 nTPM (limit 2); choroid plexus: 15.4 nTPM (limit 2); duodenum: 2.9 nTPM (limit 2); heart muscle: 5 nTPM (limit ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 47/50 (94.0%); Lung Adenocarcinoma (validation): 96/105 (91.4%); Lung Squamous Cell Carcinoma (validation): 59/68 (86.8%)

Cancer IHC. skin cancer: 10/11 (90.9%); lymphoma: 9/11 (81.8%)

Biology. May be involved in transcriptional regulation. May have a role in embryonic development

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P17027 | HPA portal (live; not the pinned values)

ZNF230 | Unsafe / RNA

ENSG00000159882

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.1 nTPM (limit 2); bone marrow: 3.2 nTPM (limit 2); cerebellum: 3.6 nTPM (limit 2); choroid plexus: 2.1 nTPM (limit 2); lung: 2.3 nTPM (limit 2); pituitary gland: 2.6 nTPM (limit 2); retina: 4.5 nTPM (limit 2); small intestine: 2.3 nTPM (limit 2); adrenal gland: 2.1 nTPM (baseline ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 44/50 (88.0%); Pancreatic Adenocarcinoma (validation): 24/80 (30.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UIE0 | HPA portal (live; not the pinned values)

ZNF233 | Unsafe / RNA

ENSG00000159915

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.3 nTPM (limit 2); cerebellum: 4.2 nTPM (limit 2); cerebral cortex: 3.1 nTPM (limit 2); heart muscle: 3 nTPM (limit 2); hippocampal formation: 2.3 nTPM (limit 2); hypothalamus: 2.7 nTPM (limit 2); pituitary gland: 3 nTPM (limit 2); retina: 4.7 nTPM (limit 2); basal ganglia: 2.3 nTPM ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 175/207 (84.5%); Colon Adenocarcinoma (validation): 390/486 (80.2%); Breast Invasive Carcinoma (validation): 38/50 (76.0%)

Cancer IHC. testis cancer: 10/10 (100.0%); lymphoma: 11/12 (91.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NK53 | HPA portal (live; not the pinned values)

ZNF257 | Unsafe / RNA

ENSG00000197134

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 4.1 nTPM (limit 2); pancreas: 2.1 nTPM (limit 2); bone marrow: 4.1 nTPM (baseline 2); pancreas: 2.1 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 32/50 (64.0%); Glioblastoma Multiforme (validation): 20/58 (34.5%); Testicular Germ Cell Tumor (TCGA): 39/133 (29.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y2Q1 | HPA portal (live; not the pinned values)

ZNF264 | Unsafe / RNA

ENSG00000083844

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.2 nTPM (limit 2); amygdala: 2.2 nTPM (limit 2); basal ganglia: 2.6 nTPM (limit 2); bone marrow: 8 nTPM (limit 2); cerebellum: 3.8 nTPM (limit 2); cerebral cortex: 3 nTPM (limit 2); choroid plexus: 3.1 nTPM (limit 2); duodenum: 4.6 nTPM (limit 2); heart muscle: 3.2 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 79/80 (98.8%); Rectum Adenocarcinoma (validation): 198/207 (95.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Curated tissue note. Relatively highly expressed in kidney, thymus, testis, ovary, brain, lung, placenta, and prostate, and relatively low expression in heart, liver, skeletal muscle, pancreas, spleen, and small intestine

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43296 | HPA portal (live; not the pinned values)

ZNF268 | Unsafe / Protein

ENSG00000090612

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 78/80 (97.5%); Lung Adenocarcinoma (validation): 102/105 (97.1%)

Cancer IHC. ovarian cancer: 12/12 (100.0%); breast cancer: 11/11 (100.0%)

Biology. Acts as a transcriptional repressor. Inhibits erythroid differentiation and tumor cell proliferation. Plays a role during ovarian cancer development and progression Contributes to cervical carcinogenesis in part through the TNF-induced NF-kappa-B signaling pathway by interacting with the I-kappa-B-kinase (IKK) core complex Involved in the regulation of antiviral interferon signaling. During viral infection, recruits SETD4 to TBK1, ... [full text in table]

Curated tissue note. Overexpressed in ovarian cancer tissues compared to normal ovarian tissues. Isoform 1 and isoform 2 are expressed in squamous epithelium tissues. Isoform 2 is overexpressed in squamous cervical cancer (at ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14587 | HPA portal (live; not the pinned values)

ZNF280B | Unsafe / RNA

ENSG00000275004

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 2.4 nTPM (limit 2); cerebral cortex: 2.6 nTPM (limit 2); hypothalamus: 3 nTPM (limit 2); retina: 3.2 nTPM (limit 2); cerebellum: 2.4 nTPM (baseline 2); cerebral cortex: 2.6 nTPM (baseline 2); hypothalamus: 3 nTPM (baseline 2); retina: 3.2 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 41/133 (30.8%); Skin Cuteneous Melanoma (TCGA): 17/99 (17.2%); Rectum Adenocarcinoma (validation): 14/207 (6.8%)

Cancer IHC. liver cancer: 2/12 (16.7%); stomach cancer: 1/11 (9.1%)

Biology. May function as a transcription factor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86YH2 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ZNF280C | Unsafe / Protein

ENSG00000056277

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Enhanced); bone marrow / hematopoietic cells: Medium (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: High (Enhanced); cerebellum / cells in granular layer: Medium (Enhanced); cerebellum / cells in molecular layer: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 206/207 (99.5%); Colon Adenocarcinoma (validation): 480/486 (98.8%)

Cancer IHC. renal cancer: 8/11 (72.7%); liver cancer: 6/11 (54.5%)

Biology. May function as a transcription factor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8ND82 | HPA portal (live; not the pinned values)

ZNF284 | Unsafe / RNA

ENSG00000186026

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.2 nTPM (limit 2); retina: 2.2 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 53/58 (91.4%); Breast Invasive Carcinoma (validation): 41/50 (82.0%); Colon Adenocarcinoma (validation): 62/486 (12.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Curated tissue note. Expressed in lung, liver, pancreas and thymus. Lower expression in heart, placenta, spleen, prostate, ovary, small intestine and colon. No expression seen in brain, skeletal muscle, kidney, testis and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q2VY69 | HPA portal (live; not the pinned values)

ZNF286A | Unsafe / RNA

ENSG00000187607

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 7.6 nTPM (limit 2); amygdala: 5.7 nTPM (limit 2); basal ganglia: 5.5 nTPM (limit 2); bone marrow: 3.9 nTPM (limit 2); cerebellum: 6.7 nTPM (limit 2); cerebral cortex: 8.1 nTPM (limit 2); choroid plexus: 7.4 nTPM (limit 2); duodenum: 3.7 nTPM (limit 2); esophagus: 9.7 nTPM (limit 6); ... [full text in table]

Cancer RNA. Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Adenocarcinoma (validation): 104/105 (99.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9HBT8 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ZNF311 | Unsafe / Protein

ENSG00000197935

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Low (Approved); caudate / glial cells: Medium (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebral cortex / endothelial cells: Low (Approved); cerebral cortex / glial cells: Medium (Approved); cerebral cortex / neuropil: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 50/58 (86.2%); Breast Invasive Carcinoma (validation): 38/50 (76.0%); Ovary Serous Cystadenocarcinoma (validation): 3/81 (3.7%)

Cancer IHC. glioma: 5/10 (50.0%); breast cancer: 0/10 (0.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5JNZ3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ZNF324B | Unsafe / RNA

ENSG00000249471

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.1 nTPM (limit 2); cerebellum: 2.7 nTPM (limit 2); cerebral cortex: 3 nTPM (limit 2); kidney: 2.2 nTPM (limit 2); liver: 2.4 nTPM (limit 2); lung: 2.2 nTPM (limit 2); pancreas: 3.5 nTPM (limit 2); pituitary gland: 3 nTPM (limit 2); skeletal muscle: 3 nTPM (limit 2); small intestine: ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 48/50 (96.0%); Rectum Adenocarcinoma (validation): 198/207 (95.7%); Colon Adenocarcinoma (validation): 459/486 (94.4%)

Cancer IHC. testis cancer: 5/9 (55.6%); liver cancer: 2/12 (16.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6AW86 | HPA portal (live; not the pinned values)

ZNF358 | Unsafe / Protein

ENSG00000198816

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Low (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / endothelial ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%)

Cancer IHC. ovarian cancer: 8/11 (72.7%); breast cancer: 8/12 (66.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NW07 | HPA portal (live; not the pinned values)

ZNF367 | Unsafe / Protein

ENSG00000165244

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in molecular layer: Medium (Uncertain); cerebellum / Purkinje cells: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 445/486 (91.6%); Breast Invasive Carcinoma (validation): 45/50 (90.0%)

Cancer IHC. glioma: 7/11 (63.6%); urothelial cancer: 5/11 (45.5%)

Biology. Transcriptional activator. Isoform 1 may be involved in transcriptional activation of erythroid genes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7RTV3 | HPA portal (live; not the pinned values)

ZNF385D | Unsafe / RNA

ENSG00000151789

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 7.2 nTPM (limit 2); cerebral cortex: 3.8 nTPM (limit 2); midbrain: 2.4 nTPM (limit 2); retina: 7.9 nTPM (limit 2); cerebellum: 7.2 nTPM (baseline 2); cerebral cortex: 3.8 nTPM (baseline 2); midbrain: 2.4 nTPM (baseline 2); retina: 7.9 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 30/50 (60.0%); Glioblastoma Multiforme (validation): 31/58 (53.4%); Glioblastoma Multiforme (TCGA): 23/141 (16.3%)

Cancer IHC. ovarian cancer: 2/11 (18.2%); cervical cancer: 1/11 (9.1%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H6B1 | HPA portal (live; not the pinned values)

ZNF391 | Unsafe / RNA

ENSG00000124613

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 3 nTPM (limit 2); cerebellum: 2.7 nTPM (limit 2); cerebral cortex: 3.1 nTPM (limit 2); hippocampal formation: 2.2 nTPM (limit 2); kidney: 2.7 nTPM (limit 2); pancreas: 2.1 nTPM (limit 2); smooth muscle: 2.3 nTPM (limit 2); basal ganglia: 3 nTPM (baseline 2); cerebellum: 2.7 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 55/58 (94.8%); Testicular Germ Cell Tumor (TCGA): 90/133 (67.7%); Breast Invasive Carcinoma (validation): 30/50 (60.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); prostate cancer: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UJN7 | HPA portal (live; not the pinned values)

ZNF395 | Unsafe / Protein

ENSG00000186918

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: High (Uncertain); cerebral cortex / glial cells: Medium (Uncertain); cerebral cortex / neuropil: Low (Uncertain); ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (TCGA): 254/254 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Lung Adenocarcinoma (validation): 105/105 (100.0%)

Cancer IHC. liver cancer: 9/12 (75.0%); breast cancer: 8/12 (66.7%)

Biology. Plays a role in papillomavirus genes transcription

Curated tissue note. Widely expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H8N7 | HPA portal (live; not the pinned values)

ZNF397 | Unsafe / Protein

ENSG00000186812

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Low ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Lung Squamous Cell Carcinoma (validation): 68/68 (100.0%)

Cancer IHC. endometrial cancer: 12/12 (100.0%); lung cancer: 12/12 (100.0%)

Biology. Isoform 3 acts as a DNA-dependent transcriptional repressor

Curated tissue note. Expressed strongly in testis, moderately in skeletal muscle, pancreas and prostate, and weakly in heart, placenta, liver, kidney, spleen, thymus and small intestine

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NF99 | HPA portal (live; not the pinned values)

ZNF407 | Unsafe / Protein

ENSG00000215421

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 483/486 (99.4%); Rectum Adenocarcinoma (validation): 203/207 (98.1%)

Cancer IHC. skin cancer: 11/12 (91.7%); colorectal cancer: 9/11 (81.8%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9C0G0 | HPA portal (live; not the pinned values)

ZNF410 | Unsafe / RNA

ENSG00000119725

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adipose tissue: 28 nTPM (limit 10); adrenal gland: 32 nTPM (limit 2); amygdala: 17 nTPM (limit 2); basal ganglia: 17.1 nTPM (limit 2); bone marrow: 57 nTPM (limit 2); cerebellum: 16.9 nTPM (limit 2); cerebral cortex: 17 nTPM (limit 2); choroid plexus: 30.7 nTPM (limit 2); colon: 29.5 nTPM (limit ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Lung Squamous Cell Carcinoma (TCGA): 489/489 (100.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcription factor that binds to the sequence motif 5'-CATCCCATAATA-3', and is specifically required to silence expression of fetal hemoglobin in adult erythroid cells (PubMed:33301730, PubMed:33859416). Prevents expression of fetal hemoglobin genes HBG1 and HBG2 through CHD4: acts as a direct transcriptional activator of CHD4, a central component of the NuRD complex that represses transcription of fetal hemoglobin genes HBG1 and ... [full text in table]

Curated tissue note. Widely expressed

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86VK4 | HPA portal (live; not the pinned values)

ZNF416 | Unsafe / RNA

ENSG00000083817

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.2 nTPM (limit 2); basal ganglia: 2.3 nTPM (limit 2); bone marrow: 2.6 nTPM (limit 2); cerebellum: 3.3 nTPM (limit 2); cerebral cortex: 2.1 nTPM (limit 2); choroid plexus: 2.5 nTPM (limit 2); heart muscle: 2.6 nTPM (limit 2); kidney: 2.5 nTPM (limit 2); lung: 2.4 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 41/50 (82.0%); Testicular Germ Cell Tumor (TCGA): 98/133 (73.7%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Acts as a mechanosensitive transcriptional activator in fibroblasts, regulating the expression of genes involved in proliferation, extracellular matrix synthesis, and contractility in response to increased matrix stiffness

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BWM5 | HPA portal (live; not the pinned values)

ZNF425 | Unsafe / RNA

ENSG00000204947

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 2.5 nTPM (limit 2); basal ganglia: 2.7 nTPM (limit 2); cerebellum: 3.4 nTPM (limit 2); cerebral cortex: 3.4 nTPM (limit 2); hippocampal formation: 2.3 nTPM (limit 2); hypothalamus: 2.3 nTPM (limit 2); kidney: 2.2 nTPM (limit 2); skeletal muscle: 3.6 nTPM (limit 2); amygdala: 2.5 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 53/58 (91.4%); Breast Invasive Carcinoma (validation): 42/50 (84.0%); Glioblastoma Multiforme (TCGA): 68/141 (48.2%)

Cancer IHC. ovarian cancer: 2/11 (18.2%); stomach cancer: 1/10 (10.0%)

Biology. Acts as a transcriptional repressor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6IV72 | HPA portal (live; not the pinned values)

ZNF43 | Unsafe / RNA

ENSG00000198521

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.9 nTPM (limit 2); amygdala: 2.7 nTPM (limit 2); basal ganglia: 2.4 nTPM (limit 2); bone marrow: 10.7 nTPM (limit 2); cerebellum: 3.7 nTPM (limit 2); cerebral cortex: 5.8 nTPM (limit 2); duodenum: 3.9 nTPM (limit 2); heart muscle: 4.8 nTPM (limit 2); hippocampal formation: 2.7 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 203/207 (98.1%); Colon Adenocarcinoma (validation): 474/486 (97.5%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May be involved in transcriptional regulation

Curated tissue note. T- and B-cell lines

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P17038 | HPA portal (live; not the pinned values)

ZNF430 | Unsafe / RNA

ENSG00000118620

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.9 nTPM (limit 2); amygdala: 2.1 nTPM (limit 2); basal ganglia: 3.4 nTPM (limit 2); bone marrow: 11.6 nTPM (limit 2); cerebellum: 4.1 nTPM (limit 2); cerebral cortex: 3.7 nTPM (limit 2); duodenum: 3.4 nTPM (limit 2); esophagus: 6.6 nTPM (limit 6); hippocampal formation: 2.2 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 205/207 (99.0%); Colon Adenocarcinoma (validation): 480/486 (98.8%)

Cancer IHC. liver cancer: 6/12 (50.0%); colorectal cancer: 3/12 (25.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H8G1 | HPA portal (live; not the pinned values)

ZNF460 | Unsafe / RNA

ENSG00000197714

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.8 nTPM (limit 2); bone marrow: 5.9 nTPM (limit 2); retina: 2.5 nTPM (limit 2); adrenal gland: 4.8 nTPM (baseline 2); bone marrow: 5.9 nTPM (baseline 2); retina: 2.5 nTPM (baseline 2)

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. prostate cancer: 9/11 (81.8%); melanoma: 8/10 (80.0%)

Biology. May be involved in transcriptional regulation

Curated tissue note. Ubiquitously expressed at low levels. Highest levels are found in pancreas and liver

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q14592 | HPA portal (live; not the pinned values)

ZNF483 | Unsafe / RNA

ENSG00000173258

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 5.8 nTPM (limit 2); basal ganglia: 2.1 nTPM (limit 2); bone marrow: 3.4 nTPM (limit 2); cerebellum: 4.2 nTPM (limit 2); cerebral cortex: 10.6 nTPM (limit 2); choroid plexus: 7.6 nTPM (limit 2); hypothalamus: 2.4 nTPM (limit 2); kidney: 2.1 nTPM (limit 2); midbrain: 2.4 nTPM (limit ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 61/80 (76.2%); Testicular Germ Cell Tumor (TCGA): 83/133 (62.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TF39 | HPA portal (live; not the pinned values)

ZNF485 | Unsafe / RNA

ENSG00000198298

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. pancreas: 4 nTPM (limit 2); skeletal muscle: 3 nTPM (limit 2); pancreas: 4 nTPM (baseline 2); skeletal muscle: 3 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Rectum Adenocarcinoma (validation): 160/207 (77.3%)

Cancer IHC. breast cancer: 1/11 (9.1%); lung cancer: 1/12 (8.3%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NCK3 | HPA portal (live; not the pinned values)

ZNF490 | Unsafe / High RNA

ENSG00000188033

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 41.6 nTPM; esophagus/plasma cells; hpa_single_cell_tissue; vital; 15.8 TPM; blood/Progenitor cell; hpa_immune_monaco; vital

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 192/207 (92.8%); Colon Adenocarcinoma (validation): 433/486 (89.1%)

Cancer IHC. colorectal cancer: 9/11 (81.8%); pancreatic cancer: 8/11 (72.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9ULM2 | HPA portal (live; not the pinned values)

ZNF492 | Unsafe / RNA

ENSG00000229676

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.5 nTPM (limit 2); bone marrow: 2.5 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 88/133 (66.2%); Glioblastoma Multiforme (validation): 11/58 (19.0%); Uterine Corpus Endometrial Carcinoma (TCGA): 13/176 (7.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P255 | HPA portal (live; not the pinned values)

ZNF525 | Unsafe / RNA

ENSG00000203326

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.5 nTPM (limit 2); amygdala: 2.1 nTPM (limit 2); basal ganglia: 2.4 nTPM (limit 2); bone marrow: 4.8 nTPM (limit 2); cerebellum: 5.7 nTPM (limit 2); choroid plexus: 2.8 nTPM (limit 2); duodenum: 3.8 nTPM (limit 2); heart muscle: 2.3 nTPM (limit 2); hippocampal formation: 2.3 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 198/207 (95.7%); Testicular Germ Cell Tumor (TCGA): 127/133 (95.5%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N782 | HPA portal (live; not the pinned values)

ZNF527 | Unsafe / RNA

ENSG00000189164

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.3 nTPM (limit 2); heart muscle: 2.2 nTPM (limit 2); kidney: 2.1 nTPM (limit 2); pituitary gland: 2.2 nTPM (limit 2); retina: 3.1 nTPM (limit 2); skeletal muscle: 2.2 nTPM (limit 2); bone marrow: 2.3 nTPM (baseline 2); heart muscle: 2.2 nTPM (baseline 2); kidney: 2.1 nTPM (baseline ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Pancreatic Adenocarcinoma (validation): 57/80 (71.2%)

Cancer IHC. ovarian cancer: 12/12 (100.0%); colorectal cancer: 11/12 (91.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NB42 | HPA portal (live; not the pinned values)

ZNF541 | Unsafe / High RNA

ENSG00000118156

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 34.7 nTPM; kidney/proximal tubular cells; hpa_single_cell_tissue; vital; 4.61 TPM; blood/TREG_NAIVE; dice_treg_naive; vital

Cancer RNA. Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 108/283 (38.2%); Kidney Renal Papillary Cell Carcinoma (TCGA): 64/282 (22.7%); Breast Invasive Carcinoma (validation): 9/50 (18.0%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/12 (0.0%)

Biology. Transcription regulator which is essential for male fertility and for the completion of meiotic prophase in spermatocytes. Regulates progression of the pachytene stage of meiotic prophase by activating the expression of genes involved in meiosis during spermatogenesis. Maintains the repression of pre-pachytene transcriptional programs, including meiotic double-strand breaks (DSB) formation genes in pachytene spermatocytes and ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H0D2 | HPA portal (live; not the pinned values)

ZNF543 | Unsafe / RNA

ENSG00000178229

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.6 nTPM (limit 2); retina: 2.6 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 74/80 (92.5%); Breast Invasive Carcinoma (validation): 44/50 (88.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q08ER8 | HPA portal (live; not the pinned values)

ZNF546 | Unsafe / RNA

ENSG00000187187

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.7 nTPM (limit 2); basal ganglia: 2.6 nTPM (limit 2); cerebellum: 2.9 nTPM (limit 2); heart muscle: 2.4 nTPM (limit 2); kidney: 2.2 nTPM (limit 2); liver: 3.6 nTPM (limit 2); pituitary gland: 2.7 nTPM (limit 2); retina: 2.3 nTPM (limit 2); skeletal muscle: 2.4 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 40/50 (80.0%); Pancreatic Adenocarcinoma (validation): 32/80 (40.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86UE3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ZNF548 | Unsafe / Protein

ENSG00000188785

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebellum / Purkinje cells: Medium (Uncertain); cerebral cortex / neuronal cells: ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Lung Squamous Cell Carcinoma (validation): 67/68 (98.5%)

Cancer IHC. breast cancer: 7/11 (63.6%); pancreatic cancer: 7/11 (63.6%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEK5 | HPA portal (live; not the pinned values)

ZNF555 | Unsafe / RNA

ENSG00000186300

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 2.7 nTPM (limit 2); cerebellum: 2.4 nTPM (limit 2); heart muscle: 2.9 nTPM (limit 2); kidney: 2.9 nTPM (limit 2); liver: 2.2 nTPM (limit 2); lung: 2.1 nTPM (limit 2); pancreas: 2.6 nTPM (limit 2); retina: 5.4 nTPM (limit 2); skeletal muscle: 2.6 nTPM (limit 2); smooth muscle: 2.1 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 47/50 (94.0%); Pancreatic Adenocarcinoma (validation): 37/80 (46.2%)

Cancer IHC. skin cancer: 6/12 (50.0%); prostate cancer: 4/8 (50.0%)

Biology. Transcriptional regulator that positively regulates SLC25A4 expression

Curated tissue note. Expressed in myoblasts (at protein level)

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8NEP9 | HPA portal (live; not the pinned values)

ZNF557 | Unsafe / RNA

ENSG00000130544

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 3.7 nTPM (limit 2); liver: 2.1 nTPM (limit 2); bone marrow: 3.7 nTPM (baseline 2); liver: 2.1 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 205/207 (99.0%); Colon Adenocarcinoma (validation): 479/486 (98.6%)

Cancer IHC. melanoma: 8/12 (66.7%); endometrial cancer: 3/11 (27.3%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N988 | HPA portal (live; not the pinned values)

ZNF559 | Unsafe / Protein

ENSG00000188321

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); melanoma: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BR84 | HPA portal (live; not the pinned values)

ZNF564 | Unsafe / RNA

ENSG00000249709

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.4 nTPM (limit 2); amygdala: 2.3 nTPM (limit 2); basal ganglia: 2.8 nTPM (limit 2); bone marrow: 4.3 nTPM (limit 2); cerebellum: 2.4 nTPM (limit 2); cerebral cortex: 2.8 nTPM (limit 2); choroid plexus: 4 nTPM (limit 2); colon: 7.2 nTPM (limit 6); duodenum: 6.1 nTPM (limit 2); heart ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%); Breast Invasive Carcinoma (validation): 49/50 (98.0%)

Cancer IHC. cervical cancer: 2/12 (16.7%); endometrial cancer: 2/12 (16.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TBZ8 | HPA portal (live; not the pinned values)

ZNF565 | Unsafe / RNA

ENSG00000196357

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.5 nTPM (limit 2); amygdala: 3.1 nTPM (limit 2); basal ganglia: 3.2 nTPM (limit 2); bone marrow: 3.6 nTPM (limit 2); cerebellum: 2.7 nTPM (limit 2); cerebral cortex: 3.5 nTPM (limit 2); choroid plexus: 3.7 nTPM (limit 2); heart muscle: 2.3 nTPM (limit 2); hippocampal formation: 3.2 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 48/50 (96.0%); Pancreatic Adenocarcinoma (validation): 48/80 (60.0%)

Cancer IHC. melanoma: 11/11 (100.0%); prostate cancer: 9/12 (75.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N9K5 | HPA portal (live; not the pinned values)

ZNF568 | Unsafe / RNA

ENSG00000198453

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.4 nTPM (limit 2); amygdala: 4.9 nTPM (limit 2); basal ganglia: 4.2 nTPM (limit 2); cerebellum: 5.9 nTPM (limit 2); cerebral cortex: 4.3 nTPM (limit 2); choroid plexus: 3.3 nTPM (limit 2); heart muscle: 3 nTPM (limit 2); hippocampal formation: 4.6 nTPM (limit 2); hypothalamus: 5.4 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 131/141 (92.9%); Breast Invasive Carcinoma (validation): 43/50 (86.0%)

Cancer IHC. renal cancer: 1/11 (9.1%); breast cancer: 0/12 (0.0%)

Biology. Has transcriptional repression activity, partially through the recruitment of the corepressor TRIM28 but also has repression activity independently of this interaction. Essential during embryonic development, where it acts as a direct repressor of a placental-specific transcript of IGF2 in early development and regulates convergent extension movements required for axis elongation and tissue morphogenesis in all germ layers. Also ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3ZCX4 | HPA portal (live; not the pinned values)

ZNF569 | Unsafe / RNA

ENSG00000196437

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4 nTPM (limit 2); amygdala: 3.5 nTPM (limit 2); basal ganglia: 3.7 nTPM (limit 2); bone marrow: 4.5 nTPM (limit 2); cerebellum: 5.5 nTPM (limit 2); cerebral cortex: 4.7 nTPM (limit 2); choroid plexus: 2.6 nTPM (limit 2); heart muscle: 4.1 nTPM (limit 2); hippocampal formation: 3.1 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Rectum Adenocarcinoma (validation): 147/207 (71.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); colorectal cancer: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5MCW4 | HPA portal (live; not the pinned values)

ZNF570 | Unsafe / Protein

ENSG00000171827

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Pancreatic Adenocarcinoma (validation): 39/80 (48.8%)

Cancer IHC. breast cancer: 11/11 (100.0%); colorectal cancer: 11/11 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96NI8 | HPA portal (live; not the pinned values)

ZNF571 | Unsafe / RNA

ENSG00000180479

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 7.3 nTPM (limit 2); amygdala: 4.4 nTPM (limit 2); basal ganglia: 3.8 nTPM (limit 2); bone marrow: 8.4 nTPM (limit 2); cerebellum: 5.2 nTPM (limit 2); cerebral cortex: 5.5 nTPM (limit 2); choroid plexus: 5.7 nTPM (limit 2); duodenum: 2.1 nTPM (limit 2); heart muscle: 4 nTPM (limit 2); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Testicular Germ Cell Tumor (TCGA): 133/133 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z3V5 | HPA portal (live; not the pinned values)

ZNF572 | Unsafe / Protein

ENSG00000180938

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / neuronal cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 37/50 (74.0%); Testicular Germ Cell Tumor (TCGA): 41/133 (30.8%)

Cancer IHC. testis cancer: 8/10 (80.0%); urothelial cancer: 9/12 (75.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7Z3I7 | HPA portal (live; not the pinned values)

ZNF573 | Unsafe / Protein

ENSG00000189144

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 53/80 (66.2%); Breast Invasive Carcinoma (validation): 26/50 (52.0%)

Cancer IHC. cervical cancer: 12/12 (100.0%); prostate cancer: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86YE8 | HPA portal (live; not the pinned values)

ZNF578 | Unsafe / RNA

ENSG00000258405

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.9 nTPM (limit 2); retina: 2.9 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 102/133 (76.7%); Glioblastoma Multiforme (validation): 34/58 (58.6%); Colon Adenocarcinoma (validation): 220/486 (45.3%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96N58 | HPA portal (live; not the pinned values)

ZNF583 | Unsafe / RNA

ENSG00000198440

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.3 nTPM (limit 2); amygdala: 3.1 nTPM (limit 2); basal ganglia: 3 nTPM (limit 2); cerebellum: 4 nTPM (limit 2); cerebral cortex: 3.7 nTPM (limit 2); hippocampal formation: 3.2 nTPM (limit 2); hypothalamus: 3 nTPM (limit 2); kidney: 2.7 nTPM (limit 2); liver: 2.5 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Glioblastoma Multiforme (TCGA): 75/141 (53.2%)

Cancer IHC. liver cancer: 5/12 (41.7%); urothelial cancer: 1/10 (10.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96ND8 | HPA portal (live; not the pinned values)

ZNF587 | Unsafe / Protein

ENSG00000198466

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Rectum Adenocarcinoma (validation): 207/207 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%)

Cancer IHC. liver cancer: 10/11 (90.9%); testis cancer: 9/12 (75.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96SQ5 | HPA portal (live; not the pinned values)

ZNF593 | Unsafe / Protein

ENSG00000142684

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Supported); adrenal gland / glandular cells: Medium (Supported); bone marrow / hematopoietic cells: High (Supported); bronchus / respiratory epithelial cells: High (Supported); caudate / glial cells: High (Supported); caudate / neuronal cells: Medium (Supported); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (TCGA): 1022/1022 (100.0%); Lung Adenocarcinoma (TCGA): 497/497 (100.0%); Thyroid Carcinoma (TCGA): 495/495 (100.0%)

Cancer IHC. breast cancer: 12/12 (100.0%); endometrial cancer: 12/12 (100.0%)

Biology. Involved in pre-60S ribosomal particles maturation by promoting the nuclear export of the 60S ribosome (PubMed:32669547). Negatively modulates the DNA binding activity of Oct-2 and therefore its transcriptional regulatory activity (PubMed:9115366)

Curated tissue note. Ubiquitous. Detected in spleen, prostate, testis, small intestine, colon and to a minor level in thymus and peripheral blood leukocytes

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O00488 | HPA portal (live; not the pinned values)

ZNF597 | Unsafe / Protein

ENSG00000167981

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. caudate / neuronal cells: Low (Approved); cerebral cortex / neuropil: Low (Approved); kidney / cells in tubules: Low (Approved)

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Pancreatic Adenocarcinoma (validation): 33/80 (41.2%)

Cancer IHC. breast cancer: 0/12 (0.0%); cervical cancer: 0/11 (0.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96LX8 | HPA portal (live; not the pinned values)

ZNF607 | Unsafe / RNA

ENSG00000198182

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.8 nTPM (limit 2); amygdala: 2.9 nTPM (limit 2); basal ganglia: 3.3 nTPM (limit 2); cerebellum: 5.7 nTPM (limit 2); cerebral cortex: 3.7 nTPM (limit 2); choroid plexus: 3.4 nTPM (limit 2); heart muscle: 2.8 nTPM (limit 2); hippocampal formation: 3.2 nTPM (limit 2); hypothalamus: 3.3 ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 47/50 (94.0%); Testicular Germ Cell Tumor (TCGA): 109/133 (82.0%)

Cancer IHC. lung cancer: 12/12 (100.0%); ovarian cancer: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96SK3 | HPA portal (live; not the pinned values)

ZNF610 | Unsafe / Protein

ENSG00000167554

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Enhanced); bronchus / respiratory epithelial cells: Medium (Enhanced); caudate / glial cells: Medium (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in granular layer: Low (Enhanced); cerebellum / cells in molecular layer: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 43/50 (86.0%); Testicular Germ Cell Tumor (TCGA): 104/133 (78.2%)

Cancer IHC. urothelial cancer: 12/12 (100.0%); pancreatic cancer: 10/11 (90.9%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N9Z0 | HPA portal (live; not the pinned values)

ZNF611 | Unsafe / RNA

ENSG00000213020

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.6 nTPM (limit 2); bone marrow: 3.4 nTPM (limit 2); cerebellum: 2.6 nTPM (limit 2); cerebral cortex: 2.8 nTPM (limit 2); duodenum: 2.3 nTPM (limit 2); heart muscle: 2.8 nTPM (limit 2); kidney: 3.8 nTPM (limit 2); lung: 2.9 nTPM (limit 2); pancreas: 4.1 nTPM (limit 2); pituitary ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 461/486 (94.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N823 | HPA portal (live; not the pinned values)

ZNF619 | Unsafe / Protein

ENSG00000177873

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 477/486 (98.1%); Rectum Adenocarcinoma (validation): 198/207 (95.7%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); melanoma: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N2I2 | HPA portal (live; not the pinned values)

ZNF620 | Unsafe / RNA

ENSG00000177842

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 3.1 nTPM (limit 2); cerebellum: 3.2 nTPM (limit 2); cerebral cortex: 2.3 nTPM (limit 2); kidney: 2.3 nTPM (limit 2); lung: 2.1 nTPM (limit 2); pancreas: 2.4 nTPM (limit 2); pituitary gland: 2.4 nTPM (limit 2); retina: 4.1 nTPM (limit 2); skeletal muscle: 2.5 nTPM (limit 2); bone ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 42/50 (84.0%); Colon Adenocarcinoma (validation): 301/486 (61.9%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); liver cancer: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZNG0 | HPA portal (live; not the pinned values)

ZNF624 | Unsafe / Protein

ENSG00000197566

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 46/50 (92.0%); Pancreatic Adenocarcinoma (validation): 22/80 (27.5%)

Cancer IHC. colorectal cancer: 9/12 (75.0%); cervical cancer: 8/11 (72.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9P2J8 | HPA portal (live; not the pinned values)

ZNF626 | Unsafe / Protein

ENSG00000188171

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebral cortex / endothelial cells: Low (Uncertain); cerebral cortex / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Kidney Renal Clear Cell Carcinoma (validation): 96/100 (96.0%); Glioblastoma Multiforme (TCGA): 133/141 (94.3%)

Cancer IHC. prostate cancer: 8/9 (88.9%); breast cancer: 9/11 (81.8%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q68DY1 | HPA portal (live; not the pinned values)

ZNF646 | Unsafe / Protein

ENSG00000167395

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Low (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebral cortex / neuronal cells: Low (Uncertain); colon / peripheral nerve/ganglion: Low (Uncertain); hippocampus / neuronal cells: Low (Uncertain); kidney / cells in glomeruli: Low (Uncertain); liver / ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 485/486 (99.8%)

Cancer IHC. testis cancer: 1/11 (9.1%); breast cancer: 0/11 (0.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O15015 | HPA portal (live; not the pinned values)

ZNF655 | Unsafe / Protein

ENSG00000197343

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (TCGA): 141/141 (100.0%); Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%)

Cancer IHC. testis cancer: 5/11 (45.5%); pancreatic cancer: 4/10 (40.0%)

Biology. Probable transcription factor

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N720 | HPA portal (live; not the pinned values)

ZNF660 | Unsafe / RNA

ENSG00000144792

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 2.3 nTPM (limit 2); pituitary gland: 2.4 nTPM (limit 2); retina: 4.4 nTPM (limit 2); cerebellum: 2.3 nTPM (baseline 2); pituitary gland: 2.4 nTPM (baseline 2); retina: 4.4 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 7/80 (8.8%); Breast Invasive Carcinoma (validation): 3/50 (6.0%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); liver cancer: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6AZW8 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ZNF668 | Unsafe / Protein

ENSG00000167394

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 50/50 (100.0%); Testicular Germ Cell Tumor (TCGA): 132/133 (99.2%); Rectum Adenocarcinoma (validation): 203/207 (98.1%)

Cancer IHC. pancreatic cancer: 8/8 (100.0%); prostate cancer: 11/12 (91.7%)

Biology. May be involved in transcriptional regulation. May play a role in DNA repair process

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96K58 | HPA portal (live; not the pinned values)

ZNF669 | Unsafe / RNA

ENSG00000188295

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 11.5 nTPM (limit 2); cerebellum: 2.9 nTPM (limit 2); cerebral cortex: 3.2 nTPM (limit 2); duodenum: 2.8 nTPM (limit 2); heart muscle: 2.4 nTPM (limit 2); kidney: 3.6 nTPM (limit 2); liver: 2.1 nTPM (limit 2); lung: 3.2 nTPM (limit 2); pancreas: 4.1 nTPM (limit 2); retina: 4.9 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 205/207 (99.0%); Colon Adenocarcinoma (validation): 477/486 (98.1%)

Cancer IHC. colorectal cancer: 9/11 (81.8%); liver cancer: 7/12 (58.3%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96BR6 | HPA portal (live; not the pinned values)

ZNF670 | Unsafe / Protein

ENSG00000277462

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Low (Uncertain); bronchus / respiratory epithelial cells: Low (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low (Uncertain); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 42/50 (84.0%); Testicular Germ Cell Tumor (TCGA): 37/133 (27.8%); Lung Squamous Cell Carcinoma (validation): 9/68 (13.2%)

Cancer IHC. testis cancer: 10/12 (83.3%); urothelial cancer: 6/12 (50.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BS34 | HPA portal (live; not the pinned values)

ZNF675 | Unsafe / RNA

ENSG00000197372

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 5.3 nTPM (limit 2); amygdala: 4.8 nTPM (limit 2); basal ganglia: 4.3 nTPM (limit 2); bone marrow: 11.2 nTPM (limit 2); cerebellum: 5 nTPM (limit 2); cerebral cortex: 10.7 nTPM (limit 2); choroid plexus: 3.8 nTPM (limit 2); colon: 8 nTPM (limit 6); duodenum: 6.5 nTPM (limit 2); heart ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 484/486 (99.6%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation. May play a role during osteoclast differentiation by modulating TRAF6 signaling activity

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8TD23 | HPA portal (live; not the pinned values)

ZNF676 | Unsafe / High RNA

ENSG00000196109

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 35.6 nTPM; brain/oligodendrocytes; hpa_single_cell_tissue; vital; 3.629 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 84/133 (63.2%); Glioblastoma Multiforme (validation): 14/58 (24.1%); Breast Invasive Carcinoma (validation): 9/50 (18.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N7Q3 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ZNF677 | Unsafe / Protein

ENSG00000197928

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / Purkinje cells: Medium (Approved); cerebral cortex / endothelial cells: Low ... [full text in table]

Cancer RNA. Prostate Adenocarcinoma (TCGA): 479/480 (99.8%); Glioblastoma Multiforme (validation): 57/58 (98.3%); Glioblastoma Multiforme (TCGA): 136/141 (96.5%)

Cancer IHC. liver cancer: 3/12 (25.0%); melanoma: 1/9 (11.1%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q86XU0 | HPA portal (live; not the pinned values)

ZNF678 | Unsafe / RNA

ENSG00000181450

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3 nTPM (limit 2); bone marrow: 3.5 nTPM (limit 2); cerebral cortex: 3.8 nTPM (limit 2); duodenum: 2.2 nTPM (limit 2); kidney: 2.5 nTPM (limit 2); liver: 2.1 nTPM (limit 2); lung: 3.1 nTPM (limit 2); pancreas: 3.2 nTPM (limit 2); retina: 5.7 nTPM (limit 2); small intestine: 2.2 nTPM ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 205/207 (99.0%); Colon Adenocarcinoma (validation): 480/486 (98.8%)

Cancer IHC. colorectal cancer: 3/11 (27.3%); pancreatic cancer: 3/12 (25.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5SXM1 | HPA portal (live; not the pinned values)

ZNF681 | Unsafe / RNA

ENSG00000196172

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.3 nTPM (limit 2); cerebral cortex: 2.3 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 46/50 (92.0%); Glioblastoma Multiforme (validation): 48/58 (82.8%); Testicular Germ Cell Tumor (TCGA): 69/133 (51.9%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96N22 | HPA portal (live; not the pinned values)

ZNF683 | Unsafe / Protein

ENSG00000176083

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: High (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Low (Uncertain); caudate / neuronal cells: High (Uncertain); cerebellum / cells in granular layer: Medium ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 34/50 (68.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 141/283 (49.8%); Lung Adenocarcinoma (TCGA): 119/497 (23.9%)

Cancer IHC. glioma: 12/12 (100.0%); urothelial cancer: 10/12 (83.3%)

Biology. Transcription factor that mediates a transcriptional program in various innate and adaptive immune tissue-resident lymphocyte T-cell types such as tissue-resident memory T (Trm), natural killer (trNK) and natural killer T (NKT) cells and negatively regulates gene expression of proteins that promote the egress of tissue-resident T-cell populations from non-lymphoid organs. Plays a role in the development, retention and long-term ... [full text in table]

Curated tissue note. Expressed in terminally differentiated effector CD8(+) T-cells, but not in naive and central memory cells (PubMed:26179882). Expressed in terminally differentiated natural killer (NK) cells and natural killer ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8IZ20 | HPA portal (live; not the pinned values)

ZNF696 | Unsafe / RNA

ENSG00000185730

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.2 nTPM (limit 2); amygdala: 4 nTPM (limit 2); basal ganglia: 4 nTPM (limit 2); bone marrow: 3.5 nTPM (limit 2); cerebellum: 5.4 nTPM (limit 2); cerebral cortex: 5.8 nTPM (limit 2); duodenum: 5.9 nTPM (limit 2); heart muscle: 2.8 nTPM (limit 2); hippocampal formation: 4.3 nTPM ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 206/207 (99.5%); Colon Adenocarcinoma (validation): 469/486 (96.5%); Breast Invasive Carcinoma (validation): 48/50 (96.0%)

Cancer IHC. colorectal cancer: 9/12 (75.0%); skin cancer: 9/12 (75.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9H7X3 | HPA portal (live; not the pinned values)

ZNF70 | Unsafe / RNA

ENSG00000187792

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 2.9 nTPM (limit 2); skeletal muscle: 2.5 nTPM (limit 2); retina: 2.9 nTPM (baseline 2); skeletal muscle: 2.5 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 45/50 (90.0%); Rectum Adenocarcinoma (validation): 162/207 (78.3%)

Cancer IHC. breast cancer: 12/12 (100.0%); prostate cancer: 11/11 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9UC06 | HPA portal (live; not the pinned values)

ZNF701 | Unsafe / RNA

ENSG00000167562

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.5 nTPM (limit 2); bone marrow: 5.3 nTPM (limit 2); duodenum: 3.2 nTPM (limit 2); heart muscle: 2.7 nTPM (limit 2); kidney: 3.5 nTPM (limit 2); lung: 3.7 nTPM (limit 2); pancreas: 2.8 nTPM (limit 2); pituitary gland: 2.2 nTPM (limit 2); retina: 3.3 nTPM (limit 2); skeletal muscle: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 206/207 (99.5%); Colon Adenocarcinoma (validation): 476/486 (97.9%)

Cancer IHC. prostate cancer: 11/12 (91.7%); urothelial cancer: 11/12 (91.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9NV72 | HPA portal (live; not the pinned values)

ZNF709 | Unsafe / RNA

ENSG00000242852

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 2.3 nTPM (limit 2); cerebellum: 2.8 nTPM (limit 2); retina: 3 nTPM (limit 2); basal ganglia: 2.3 nTPM (baseline 2); cerebellum: 2.8 nTPM (baseline 2); retina: 3 nTPM (baseline 2)

Cancer RNA. Breast Invasive Carcinoma (validation): 39/50 (78.0%); Testicular Germ Cell Tumor (TCGA): 24/133 (18.0%); Ovary Serous Cystadenocarcinoma (TCGA): 49/349 (14.0%)

Cancer IHC. colorectal cancer: 11/11 (100.0%); skin cancer: 11/11 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N972 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ZNF711 | Unsafe / Protein

ENSG00000147180

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Enhanced); caudate / glial cells: Low (Enhanced); caudate / neuronal cells: Medium (Enhanced); cerebellum / cells in molecular layer: Low (Enhanced); cerebellum / Purkinje cells: Low (Enhanced); cerebral cortex / glial cells: High (Enhanced); cerebral cortex / ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 120/141 (85.1%); Breast Invasive Carcinoma (validation): 40/50 (80.0%)

Cancer IHC. glioma: 7/10 (70.0%); skin cancer: 2/9 (22.2%)

Biology. Transcription regulator required for brain development (PubMed:20346720). Probably acts as a transcription factor that binds to the promoter of target genes and recruits PHF8 histone demethylase, leading to activated expression of genes involved in neuron development, such as KDM5C (PubMed:20346720, PubMed:31691806). May compete with transcription factor ARX for activation of expression of KDM5C (PubMed:31691806)

Curated tissue note. Expressed in neural tissues

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9Y462 | HPA portal (live; not the pinned values)

ZNF713 | Unsafe / RNA

ENSG00000178665

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebral cortex: 2.3 nTPM (limit 2); retina: 2.3 nTPM (limit 2); spinal cord: 2.7 nTPM (limit 2); cerebral cortex: 2.3 nTPM (baseline 2); retina: 2.3 nTPM (baseline 2); spinal cord: 2.7 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 48/50 (96.0%); Pancreatic Adenocarcinoma (validation): 29/80 (36.2%)

Cancer IHC. melanoma: 2/11 (18.2%); testis cancer: 1/10 (10.0%)

Biology. May be involved in transcriptional regulation

Curated tissue note. Expressed in fetal and adult brain

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q8N859 | HPA portal (live; not the pinned values)

ZNF714 | Unsafe / RNA

ENSG00000160352

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 4.3 nTPM (limit 2); basal ganglia: 3.1 nTPM (limit 2); bone marrow: 6.8 nTPM (limit 2); cerebellum: 6.3 nTPM (limit 2); cerebral cortex: 3.9 nTPM (limit 2); duodenum: 2.4 nTPM (limit 2); pancreas: 3.7 nTPM (limit 2); retina: 3.1 nTPM (limit 2); skeletal muscle: 2.7 nTPM (limit 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 409/486 (84.2%); Rectum Adenocarcinoma (validation): 166/207 (80.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

HPA portal (live; not the pinned values)

ZNF717 | Unsafe / RNA

ENSG00000227124

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 5.6 nTPM (limit 2); amygdala: 4.4 nTPM (limit 2); basal ganglia: 5.1 nTPM (limit 2); bone marrow: 2.7 nTPM (limit 2); cerebellum: 4.1 nTPM (limit 2); cerebral cortex: 4.3 nTPM (limit 2); choroid plexus: 4 nTPM (limit 2); duodenum: 5.5 nTPM (limit 2); heart muscle: 5.7 nTPM (limit 2); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 205/207 (99.0%); Glioblastoma Multiforme (validation): 56/58 (96.6%); Colon Adenocarcinoma (validation): 463/486 (95.3%)

Cancer IHC. colorectal cancer: 6/12 (50.0%); ovarian cancer: 5/12 (41.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BY31 | HPA portal (live; not the pinned values)

ZNF718 | Unsafe / RNA

ENSG00000250312

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.9 nTPM (limit 2); bone marrow: 2.9 nTPM (limit 2); cerebellum: 2.1 nTPM (limit 2); cerebral cortex: 2.8 nTPM (limit 2); kidney: 2.4 nTPM (limit 2); pancreas: 2.5 nTPM (limit 2); retina: 2.6 nTPM (limit 2); adrenal gland: 2.9 nTPM (baseline 2); bone marrow: 2.9 nTPM (baseline 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Breast Invasive Carcinoma (validation): 47/50 (94.0%); Pancreatic Adenocarcinoma (validation): 59/80 (73.8%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3SXZ3 | HPA portal (live; not the pinned values)

ZNF749 | Unsafe / RNA

ENSG00000186230

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. cerebellum: 2.3 nTPM (limit 2); retina: 3.9 nTPM (limit 2); cerebellum: 2.3 nTPM (baseline 2); retina: 3.9 nTPM (baseline 2)

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 414/486 (85.2%); Breast Invasive Carcinoma (validation): 41/50 (82.0%)

Cancer IHC. liver cancer: 5/12 (41.7%); colorectal cancer: 3/11 (27.3%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt O43361 | HPA portal (live; not the pinned values)

ZNF763 | Unsafe / Protein

ENSG00000197054

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); cerebral cortex / endothelial cells: Low (Approved); cerebral cortex / glial cells: Medium (Approved); colon / endothelial cells: Low ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 45/50 (90.0%); Glioblastoma Multiforme (validation): 28/58 (48.3%); Kidney Chromophobe (TCGA): 28/64 (43.8%)

Cancer IHC. colorectal cancer: 4/11 (36.4%); liver cancer: 3/11 (27.3%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q0D2J5 | HPA portal (live; not the pinned values)

ZNF765 | Unsafe / RNA

ENSG00000196417

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 3.7 nTPM (limit 2); bone marrow: 4.1 nTPM (limit 2); duodenum: 3 nTPM (limit 2); kidney: 2.7 nTPM (limit 2); lung: 3 nTPM (limit 2); pancreas: 4.4 nTPM (limit 2); parathyroid gland: 8.5 nTPM (limit 6); small intestine: 2.7 nTPM (limit 2); smooth muscle: 2.1 nTPM (limit 2); adrenal ... [full text in table]

Cancer RNA. Pancreatic Adenocarcinoma (validation): 80/80 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 482/486 (99.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q7L2R6 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ZNF774 | Unsafe / RNA

ENSG00000196391

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 2.6 nTPM (limit 2); amygdala: 2.2 nTPM (limit 2); basal ganglia: 3.5 nTPM (limit 2); cerebellum: 5.1 nTPM (limit 2); cerebral cortex: 4.6 nTPM (limit 2); duodenum: 2.3 nTPM (limit 2); heart muscle: 2.7 nTPM (limit 2); hypothalamus: 3 nTPM (limit 2); midbrain: 2.2 nTPM (limit 2); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 203/207 (98.1%); Colon Adenocarcinoma (validation): 468/486 (96.3%); Breast Invasive Carcinoma (validation): 34/50 (68.0%)

Cancer IHC. glioma: 10/11 (90.9%); renal cancer: 2/11 (18.2%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6NX45 | HPA portal (live; not the pinned values)

ZNF776 | Unsafe / RNA

ENSG00000152443

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. adrenal gland: 6.3 nTPM (limit 2); amygdala: 5.5 nTPM (limit 2); basal ganglia: 5.9 nTPM (limit 2); bone marrow: 16.9 nTPM (limit 2); cerebellum: 14.2 nTPM (limit 2); cerebral cortex: 6.3 nTPM (limit 2); choroid plexus: 6 nTPM (limit 2); colon: 6.2 nTPM (limit 6); duodenum: 6.2 nTPM (limit 2); ... [full text in table]

Cancer RNA. Rectum Adenocarcinoma (validation): 207/207 (100.0%); Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 483/486 (99.4%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); liver cancer: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q68DI1 | HPA portal (live; not the pinned values)

ZNF778 | Unsafe / Protein

ENSG00000170100

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in molecular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 44/50 (88.0%); Testicular Germ Cell Tumor (TCGA): 52/133 (39.1%)

Cancer IHC. glioma: 12/12 (100.0%); lung cancer: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96MU6 | HPA portal (live; not the pinned values)

ZNF782 | Unsafe / Protein

ENSG00000196597

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Colon Adenocarcinoma (validation): 470/486 (96.7%); Rectum Adenocarcinoma (validation): 198/207 (95.7%)

Cancer IHC. prostate cancer: 12/12 (100.0%); stomach cancer: 11/12 (91.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6ZMW2 | HPA portal (live; not the pinned values)

ZNF790 | Unsafe / Protein

ENSG00000197863

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / Purkinje cells: High ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Glioblastoma Multiforme (TCGA): 123/141 (87.2%); Kidney Renal Clear Cell Carcinoma (validation): 83/100 (83.0%)

Cancer IHC. breast cancer: 11/11 (100.0%); prostate cancer: 10/10 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6PG37 | HPA portal (live; not the pinned values)

ZNF799 | Unsafe / Protein

ENSG00000196466

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Approved); adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Breast Invasive Carcinoma (validation): 47/50 (94.0%); Pancreatic Adenocarcinoma (validation): 57/80 (71.2%)

Cancer IHC. breast cancer: 12/12 (100.0%); pancreatic cancer: 11/11 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96GE5 | HPA portal (live; not the pinned values)

ZNF805 | Unsafe / Protein

ENSG00000204524

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: Medium (Uncertain); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 79/80 (98.8%); Colon Adenocarcinoma (validation): 479/486 (98.6%)

Cancer IHC. prostate cancer: 10/11 (90.9%); breast cancer: 6/11 (54.5%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5CZA5 | HPA portal (live; not the pinned values)

ZNF81 | Unsafe / Protein

ENSG00000197779

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: High (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: High (Approved); cerebellum / cells in molecular layer: Low (Approved); cerebellum / Purkinje cells: Medium ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 196/207 (94.7%); Colon Adenocarcinoma (validation): 451/486 (92.8%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); ovarian cancer: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P51508 | HPA portal (live; not the pinned values)

ZNF845 | Unsafe / RNA

ENSG00000213799

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 4.9 nTPM (limit 2); duodenum: 2.4 nTPM (limit 2); kidney: 2.4 nTPM (limit 2); lung: 2.8 nTPM (limit 2); small intestine: 2.5 nTPM (limit 2); bone marrow: 4.9 nTPM (baseline 2); duodenum: 2.4 nTPM (baseline 2); kidney: 2.4 nTPM (baseline 2); lung: 2.8 nTPM (baseline 2); small intestine: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Pancreatic Adenocarcinoma (validation): 79/80 (98.8%); Breast Invasive Carcinoma (validation): 47/50 (94.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q96IR2 | HPA portal (live; not the pinned values)

ZNF846 | Unsafe / Protein

ENSG00000196605

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Low (Approved); cerebral cortex / endothelial cells: Medium (Approved); colon / endothelial cells: Medium (Approved); colon / peripheral nerve/ganglion: Low (Approved); esophagus / squamous epithelial cells: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Rectum Adenocarcinoma (validation): 204/207 (98.6%); Colon Adenocarcinoma (validation): 455/486 (93.6%)

Cancer IHC. liver cancer: 1/11 (9.1%); breast cancer: 0/11 (0.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q147U1 | HPA portal (live; not the pinned values)

ZNF85 | Unsafe / Protein

ENSG00000105750

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Approved); adrenal gland / glandular cells: High (Approved); bone marrow / hematopoietic cells: Medium (Approved); bronchus / respiratory epithelial cells: High (Approved); caudate / glial cells: Low (Approved); caudate / neuronal cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 44/50 (88.0%); Testicular Germ Cell Tumor (TCGA): 96/133 (72.2%)

Cancer IHC. testis cancer: 11/12 (91.7%); pancreatic cancer: 10/11 (90.9%)

Biology. May be a transcriptional repressor

Curated tissue note. Widely expressed, including in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q03923 | HPA portal (live; not the pinned values)

Shared UniProt entry: annotation is not independently proven for each encoding locus.

ZNF879 | Unsafe / Protein

ENSG00000234284

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Medium (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Low (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); caudate / neuronal cells: Low (Uncertain); cerebellum / cells in granular layer: ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 40/50 (80.0%); Testicular Germ Cell Tumor (TCGA): 49/133 (36.8%)

Cancer IHC. colorectal cancer: 10/10 (100.0%); lung cancer: 11/12 (91.7%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt B4DU55 | HPA portal (live; not the pinned values)

ZNF93 | Unsafe / RNA

ENSG00000184635

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. bone marrow: 3 nTPM (limit 2); cerebral cortex: 2.1 nTPM (limit 2); pancreas: 3 nTPM (limit 2); retina: 3.2 nTPM (limit 2); smooth muscle: 3.8 nTPM (limit 2); bone marrow: 3 nTPM (baseline 2); cerebral cortex: 2.1 nTPM (baseline 2); pancreas: 3 nTPM (baseline 2); retina: 3.2 nTPM (baseline 2); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Testicular Germ Cell Tumor (TCGA): 128/133 (96.2%); Rectum Adenocarcinoma (validation): 185/207 (89.4%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. Transcription factor specifically required to repress long interspersed nuclear element 1 (L1) retrotransposons: recognizes and binds L1 sequences and repress their expression by recruiting a repressive complex containing TRIM28/KAP1 (PubMed:25274305). Not able to repress expression of all subtypes of L1 elements. Binds to the 5' end of L1PA4, L1PA5 and L1PA6 subtypes, and some L1PA3 subtypes. Does not bind to L1PA7 or older subtypes ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P35789 | HPA portal (live; not the pinned values)

ZNF98 | Unsafe / RNA

ENSG00000197360

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. basal ganglia: 5 nTPM (limit 2); cerebral cortex: 3.2 nTPM (limit 2); pancreas: 35.9 nTPM (limit 2); basal ganglia: 5 nTPM (baseline 2); cerebral cortex: 3.2 nTPM (baseline 2); pancreas: 35.9 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 79/133 (59.4%); Breast Invasive Carcinoma (validation): 14/50 (28.0%); Kidney Chromophobe (TCGA): 16/64 (25.0%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NK75 | HPA portal (live; not the pinned values)

ZNG1A | Unsafe / Protein

ENSG00000172785

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bone marrow / hematopoietic cells: Low (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: Medium (Approved); cerebellum / cells in granular layer: Low ... [full text in table]

Cancer RNA. Colon Adenocarcinoma (validation): 486/486 (100.0%); Prostate Adenocarcinoma (TCGA): 480/480 (100.0%); Colon Adenocarcinoma (TCGA): 254/254 (100.0%)

Cancer IHC. prostate cancer: 12/12 (100.0%); pancreatic cancer: 11/11 (100.0%)

Biology. Zinc chaperone that directly transfers zinc cofactor to target metalloproteins, thereby activating them. Catalyzes zinc insertion into the active site of methionine aminopeptidase METAP1, which function to cleave the initiator methionine from polypeptides during or after protein translation. Mechanistically, the N-terminal psi-PxLVp motif binds to the C6H2-type zinc finger of inactive form of METAP1. After formation of the docked ... [full text in table]

Curated tissue note. Ubiquitously expressed. Up-regulated in cultured astrocytes treated with dopamine

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BRT8 | HPA portal (live; not the pinned values)

ZNG1F | Unsafe / Protein

ENSG00000215126

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Approved); bronchus / respiratory epithelial cells: Medium (Approved); caudate / glial cells: Medium (Approved); caudate / neuronal cells: High (Approved); cerebellum / cells in granular layer: Low (Approved); cerebellum / Purkinje cells: Low (Approved); ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Pancreatic Adenocarcinoma (validation): 46/80 (57.5%); Lung Squamous Cell Carcinoma (validation): 11/68 (16.2%)

Cancer IHC. colorectal cancer: 12/12 (100.0%); breast cancer: 11/11 (100.0%)

Biology. Zinc chaperone that directly transfers zinc cofactor to target metalloproteins, thereby activating them (By similarity). Catalyzes zinc insertion into the active site of methionine aminopeptidase METAP1, which function to cleave the initiator methionine from polypeptides during or after protein translation (PubMed:35584702). Mechanistically, the N-terminal psi-PxLVp motif binds to the C6H2-type zinc finger of inactive form of METAP1 ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q4V339 | HPA portal (live; not the pinned values)

ZPBP | Unsafe / RNA

ENSG00000042813

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. retina: 4.5 nTPM (limit 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%); Kidney Renal Papillary Cell Carcinoma (TCGA): 2/282 (0.7%); Uterine Corpus Endometrial Carcinoma (TCGA): 1/176 (0.6%)

Cancer IHC. glioma: 2/10 (20.0%); breast cancer: 0/12 (0.0%)

Biology. Plays a role in acrosome compaction and sperm morphogenesis (PubMed:21911476). Is implicated in sperm-oocyte interaction during fertilization (By similarity)

Curated tissue note. Expressed specifically in testis

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BS86 | HPA portal (live; not the pinned values)

ZRANB3 | Unsafe / High RNA

ENSG00000121988

Decision. At least one non-permitted or unsited RNA observation is >30 TPM or nTPM.

Normal RNA / IHC. 89.5 nTPM; brain/excitatory neurons; hpa_single_cell_tissue; vital; 8.115 TPM; blood/B_CELL_NAIVE; dice_b_cell_naive; vital

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 41/50 (82.0%); Pancreatic Adenocarcinoma (validation): 29/80 (36.2%)

Cancer IHC. No exact-ID IHC coverage in the HPA cancer snapshot

Biology. DNA annealing helicase and endonuclease required to maintain genome stability at stalled or collapsed replication forks by facilitating fork restart and limiting inappropriate recombination that could occur during template switching events (PubMed:21078962, PubMed:22704558, PubMed:22705370, PubMed:22759634, PubMed:26884333). Recruited to the sites of stalled DNA replication by polyubiquitinated PCNA and acts as a structure-specific ... [full text in table]

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q5FWF4 | HPA portal (live; not the pinned values)

ZSCAN20 | Unsafe / Protein

ENSG00000121903

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: High (Uncertain); adrenal gland / glandular cells: High (Uncertain); bone marrow / hematopoietic cells: High (Uncertain); bronchus / respiratory epithelial cells: High (Uncertain); caudate / glial cells: Medium (Uncertain); caudate / neuronal cells: High (Uncertain); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 57/58 (98.3%); Breast Invasive Carcinoma (validation): 40/50 (80.0%); Rectum Adenocarcinoma (validation): 70/207 (33.8%)

Cancer IHC. breast cancer: 12/12 (100.0%); cervical cancer: 12/12 (100.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P17040 | HPA portal (live; not the pinned values)

ZSCAN23 | Unsafe / Protein

ENSG00000187987

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adipose tissue / adipocytes: Low (Uncertain); adrenal gland / glandular cells: Medium (Uncertain); bone marrow / hematopoietic cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); cerebellum / cells in granular layer: Medium (Uncertain); cerebral cortex / glial ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 56/58 (96.6%); Breast Invasive Carcinoma (validation): 24/50 (48.0%); Ovary Serous Cystadenocarcinoma (validation): 1/81 (1.2%)

Cancer IHC. ovarian cancer: 6/11 (54.5%); breast cancer: 6/12 (50.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q3MJ62 | HPA portal (live; not the pinned values)

ZSCAN5A | Unsafe / Protein

ENSG00000131848

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. adrenal gland / glandular cells: Medium (Uncertain); bronchus / respiratory epithelial cells: Medium (Uncertain); colon / glandular cells: Medium (Uncertain); duodenum / glandular cells: Medium (Uncertain); kidney / cells in tubules: Medium (Uncertain); nasopharynx / respiratory epithelial cells: ... [full text in table]

Cancer RNA. Breast Invasive Carcinoma (validation): 43/50 (86.0%); Glioblastoma Multiforme (TCGA): 103/141 (73.0%); Glioblastoma Multiforme (validation): 40/58 (69.0%)

Cancer IHC. stomach cancer: 1/9 (11.1%); testis cancer: 1/10 (10.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q9BUG6 | HPA portal (live; not the pinned values)

ZSCAN5B | Unsafe / Protein

ENSG00000197213

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. cerebral cortex / neuronal cells: Low (Uncertain); colon / glandular cells: High (Uncertain); duodenum / glandular cells: Medium (Uncertain); kidney / cells in tubules: High (Uncertain); liver / cholangiocytes: Low (Uncertain); pancreas / exocrine glandular cells: Medium (Uncertain); rectum / ... [full text in table]

Cancer RNA. Bladder Urothelial Carcinoma (TCGA): 3/169 (1.8%); Liver Hepatocellular Carcinoma (validation): 4/231 (1.7%); Testicular Germ Cell Tumor (TCGA): 2/133 (1.5%)

Cancer IHC. colorectal cancer: 9/12 (75.0%); stomach cancer: 6/12 (50.0%)

Biology. May be involved in transcriptional regulation

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt A6NJL1 | HPA portal (live; not the pinned values)

ZXDA | Unsafe / Protein

ENSG00000198205

Decision. Normal IHC is positive outside permitted compartments.

Normal RNA / IHC. bronchus / respiratory epithelial cells: Medium (Approved); caudate / neuronal cells: Low (Approved); cerebellum / cells in granular layer: Medium (Approved); cerebellum / Purkinje cells: Low (Approved); cerebral cortex / neuronal cells: Low (Approved); colon / glandular cells: Medium (Approved); ... [full text in table]

Cancer RNA. Glioblastoma Multiforme (validation): 58/58 (100.0%); Breast Invasive Carcinoma (validation): 42/50 (84.0%); Rectum Adenocarcinoma (validation): 121/207 (58.5%)

Cancer IHC. prostate cancer: 12/12 (100.0%); stomach cancer: 12/12 (100.0%)

Biology. Cooperates with CIITA to promote transcription of MHC class I and MHC class II genes

Curated tissue note. May be expressed in brain, heart, kidney, liver, lung, muscle and placenta

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt P98168 | HPA portal (live; not the pinned values)

ZYG11A | Unsafe / RNA

ENSG00000203995

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. kidney: 2.8 nTPM (limit 2); kidney: 2.8 nTPM (baseline 2)

Cancer RNA. Testicular Germ Cell Tumor (TCGA): 107/133 (80.5%); Breast Invasive Carcinoma (validation): 36/50 (72.0%); Cervical Squamous Cell Carcinoma and Endocervical Adenocarcinoma (TCGA): 46/283 (16.3%)

Cancer IHC. stomach cancer: 9/10 (90.0%); colorectal cancer: 7/12 (58.3%)

Biology. Probably acts as a target recruitment subunit in an E3 ubiquitin ligase complex ZYGA-CUL2-elongin BC

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

UniProt Q6WRX3 | HPA portal (live; not the pinned values)

ENSG00000187186 | Unsafe / RNA

ENSG00000187186

Decision. Normal bulk RNA exceeds a category/IHC-dependent ceiling.

Normal RNA / IHC. amygdala: 57.3 nTPM (limit 2); basal ganglia: 95.6 nTPM (limit 2); cerebellum: 33 nTPM (limit 12); cerebral cortex: 75.4 nTPM (limit 12); choroid plexus: 28.6 nTPM (limit 2); heart muscle: 27 nTPM (limit 12); hippocampal formation: 56.4 nTPM (limit 2); hypothalamus: 47.3 nTPM (limit 2); kidney: ... [full text in table]

Cancer RNA. Thyroid Carcinoma (TCGA): 495/495 (100.0%); Pancreatic Adenocarcinoma (TCGA): 166/176 (94.3%); Prostate Adenocarcinoma (TCGA): 449/480 (93.5%)

Cancer IHC. stomach cancer: 2/11 (18.2%); urothelial cancer: 2/11 (18.2%)

Biology. No functional description is linked by an exact Ensembl cross-reference in this reviewed UniProt snapshot.

Therapeutic evidence. No trial link in the curated intake; not an exhaustive negative search.

HPA portal (live; not the pinned values)