

Anti- SLC6A1 Antibody HMV4721 / Recombinant Rabbit monoclonal

Human SwissProt	P31648
Human Gene Symbol	SLC6A1
Synonyms	solute carrier family 6 (neurotransmitter transporter, GABA), member 1 , A730043E01 , GABATHG , GABATR , GAT-1 , Gabt , Gabt1 , Gat1 , XT-1 , Xtrp1
Specificity	SLC6A1
Immunogen	Recombinant human SLC6A1 fragment
Isotype	Rabbit / IgG
Species Reactivity	Human

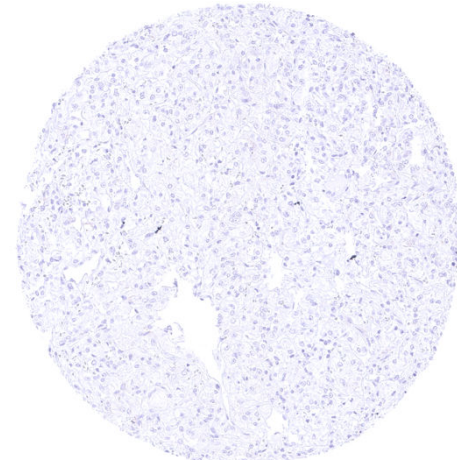
Localization	Membranous
Storage & Stability	Antibody with azide – store at 2 to 8 C. Antibody without azide – store at -20 to -80 C. Antibody is stable for 24 months. Non-hazardous. No MSD required.
Supplied As	Purified antibody from Bioreactor Concentrate by Protein A/G. Prepared in 10mM PBS with <1% BSA & <0.1% azide. Antibody concentrate is optimized for dilution within dilution range using commercially available antibody diluent for IHC.
Positive Control	Liver: A variable, weak to strong, membranous SLC6A1 staining must be seen in hepatocytes.
Negative Control	Lymph node: SLC6A1 staining must be absent in all cell types.



Cerebellum with a strong membranous SLC6A1 staining of Purkinje cells, other neurons and axons.



Liver with a moderate to strong, membranous SLC6A1 staining of hepatocytes.



SLC6A1 negative clear cell carcinoma.

Biology

Solute carrier family 6 member 1 (SLC6A1) gene a sodium-dependent neurotransmitter transporter for glycine. It plays a crucial role in the regulation of synaptic transmission and modulation of excitatory signals within the central nervous system. Accordingly, SLC6A1 is primarily expressed in the brain, notably in the spinal cord, cerebellum, and various regions of the cerebral cortex. Germline mutations have been linked to neurodevelopmental disorders, particularly epilepsy syndromes, such as severe myoclonic epilepsy in infancy. SLC6A1-associated neurodevelopmental disorders are among the top ten most common monogenic causes for both autism and epilepsy. Several SLC6A1 variants exhibit reduced transmembranous trafficking which might be rescuable by pharmacochaperoning. SLC6A1 is also found in several extra-cranial tissues like the liver, the thymus and the parathyroid, indicating a broader physiological role. Among cancers, SLC6A1 expression is particularly frequent in hepatocellular carcinoma.

Potential Research Applications

- How do different pathogenic SLC6A1 variants alter SLC6A1 trafficking, localization, and functional expression in human neurons?
- What are the cell-type specific SLC6A1 expression patterns and developmental trajectories of SLC6A1 in human brain regions implicated in epilepsy and neurodevelopmental disorders?

- How does altered SLC6A1 function affect GABAergic synapse physiology, network excitability, and seizure susceptibility in human-relevant models?
- Can pharmacological chaperones or gene therapy restore mutant SLC6A1 function, and what are optimal delivery strategies and windows for therapeutic rescue?
- What is the role of SLC6A1 in the normal liver and in liver cancer?

Protocol Suggestions

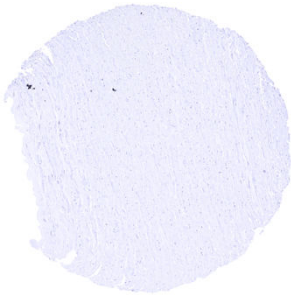
Dilution: 1:100 - 1:200; pH 7,8 is optimal. Freshly cut sections should be used (more than 10 days between cutting and staining deteriorates staining intensity for most antibodies in IHC).

Limitations

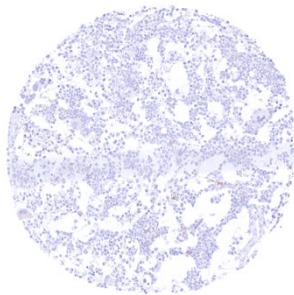
This antibody is available for **research use only** and is not approved for use in diagnostics.

Warranty

There are no warranties, expressed or implied, which extend beyond this description. MSVA is not liable for any personal injury or economic loss resulting from this product.



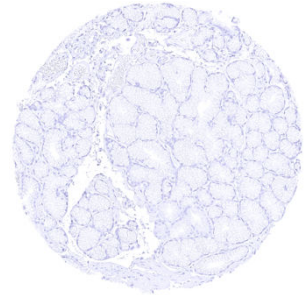
Aorta, media



Bone marrow



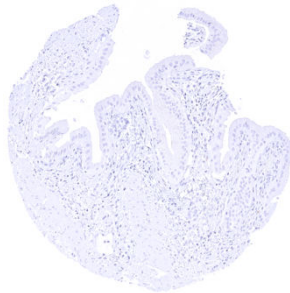
Cerebellum (white matter) – Strong SLC6A1 staining of axons.



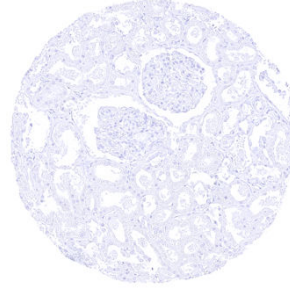
Duodenum, Brunner gland



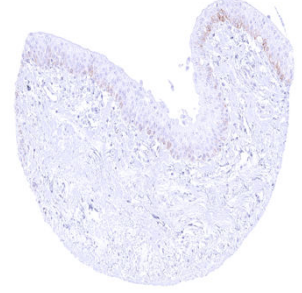
Esophagus, squamous epithelium – Weak to moderate membranous SLC6A1 staining of basal and some suprabasal squamous epithelial cells.



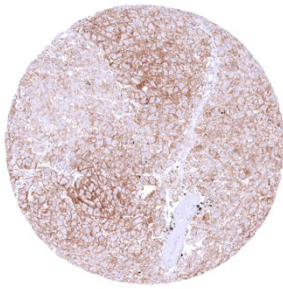
Gallbladder, epithelium



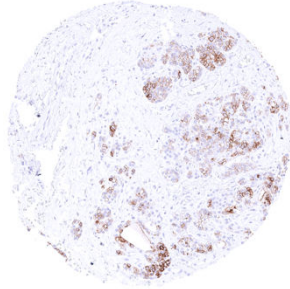
Kidney, cortex – Absence of SLC6A1 staining in this sample.



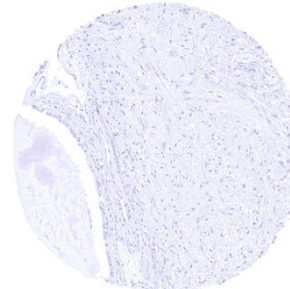
Kidney, urothelium – Weak to moderate membranous SLC6A1 staining of a subset of basal urothelial cells in this sample.



Liver – Membranous SLC6A1 staining of variable intensity of hepatocytes ranging from absent-weak to strong.



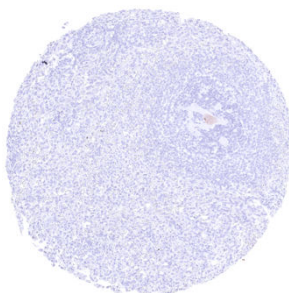
Parathyroid gland – Moderate to strong membranous staining of a subset of epithelial cells.



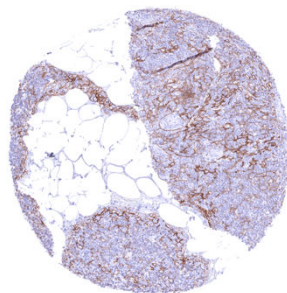
Pituitary gland, posterior lobe



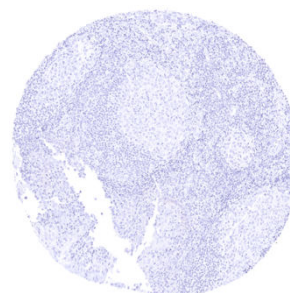
Skeletal muscle – Cytoplasmic SLC6A1 staining of a subset of skeletal muscle fibers.



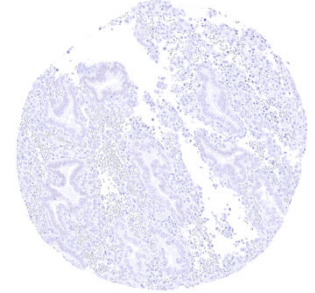
Spleen



Thymus – Strong membranous SLC6A1 staining of most if not all thymic epithelial cells.



Tonsil



Uterus, endometrium (secretion)