SLC1A3 gene

solute carrier family 1 member 3

Normal Function

The *SLC1A3* gene provides instructions for making a protein called excitatory amino acid transporter 1 (EAAT1). The EAAT1 protein is primarily found in nervous system cells called glia (or glial cells), which protect and maintain nerve cells (neurons). While the protein is found in cells throughout the brain, it is most abundant in the part of the brain that is connected to the spinal cord (the brainstem) and the region of the brain involved in coordinating movements (the cerebellum).

The EAAT1 protein transports a molecule called glutamate in the brain. Glutamate is one of several brain chemicals called neurotransmitters, which allow neurons to communicate with one another. EAAT1 also transports negatively charged chlorine atoms (chloride ions).

Neurotransmitters (such as glutamate) are released from neurons and relay signals to other cells by attaching to receptor proteins on neighboring neurons. After the neurotransmitters have had their effect, they detach from their receptors and must be cleared from the spaces between neurons. Researchers have determined that EAAT1 is one of several glutamate transporters that clear excess glutamate from these spaces. This process is carefully regulated to ensure that signals are transmitted accurately throughout the nervous system. The timely removal of glutamate is also necessary to prevent a buildup of this neurotransmitter between cells, which would be toxic to neurons.

The EAAT1 protein also transports chloride ions in and out of cells. This function of the protein may help regulate the size (volume) of glia by playing a role in their intake and release of water. EAAT1 may also help maintain a normal balance of charged molecules (ions) in cells.

Health Conditions Related to Genetic Changes

Episodic ataxia

Several variants (also called mutations) in the *SLC1A3* gene have been found to cause episodic ataxia type 6 (EA6), which is a nervous system disorder involving episodes of poor coordination and balance (ataxia).

The variants that cause episodic ataxia change single protein building blocks (amino acids) in the EAAT1 protein. Research shows that the altered EAAT1 proteins allow more chloride ions to flow in or out of cells than usual. Researchers suspect that glia with these altered channels cannot maintain the correct volume or ion balance, causing them to shrink and die.

The loss of glia is thought to impair the clearance of glutamate from between neurons in the brain. The excess glutamate may overexcite certain neurons, disrupting normal communication between these cells. The changes in signaling between neurons likely causes the episodes of uncoordinated movement seen in people with episodic ataxia.

Other Names for This Gene

- EA6
- EAA1_HUMAN
- EAAT-1
- EAAT1
- Excitatory amino acid transporter 1
- FLJ25094
- GLAST
- GLAST1
- Glial high affinity glutamate transporter
- Glutamate/aspartate transporter, high affinity, sodium-dependent
- Sodium-dependent glutamate/aspartate transporter 1
- solute carrier family 1 (glial high affinity glutamate transporter), member 3
- Solute carrier family 1 member 3

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLC1A3 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6507[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SLC1A3%5BTIAB%5D%29+OR+%28%28EAAT1%5BTIAB%5D%29+OR+%28GLAST%5BTIAB%5D%29+OR+%28GLAST1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 1 (GLIAL HIGH AFFINITY GLUTAMATE TRANSPORTER), MEMBER 3; SLC1A3 (https://omim.org/entry/600111)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6507)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SLC1A3[gene])

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Genomic Location

The *SLC1A3* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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