SLC2A1 gene

solute carrier family 2 member 1

Normal Function

The *SLC2A1* gene provides instructions for producing a protein called the glucose transporter protein type 1 (GLUT1). The GLUT1 protein is embedded in the outer membrane surrounding cells, where it transports a simple sugar called glucose into cells from the blood or from other cells for use as fuel.

In the brain, the GLUT1 protein is involved in moving glucose, which is the brain's main energy source, across the blood-brain barrier. The blood-brain barrier acts as a boundary between tiny blood vessels (capillaries) and the surrounding brain tissue; it protects the brain's delicate nerve tissue by preventing many other types of molecules from entering the brain. The GLUT1 protein also moves glucose between cells in the brain called glia, which protect and maintain nerve cells (neurons).

Health Conditions Related to Genetic Changes

GLUT1 deficiency syndrome

More than 150 *SLC2A1* gene mutations have been reported in people with GLUT1 deficiency syndrome. This disorder leads to a variety of neurological symptoms that can include developmental delay, intellectual disability, movement problems, and frequent seizures (epilepsy). The mutations that cause GLUT1 deficiency syndrome reduce or eliminate the function of the GLUT1 protein. Having less functional GLUT1 protein reduces the amount of glucose available to brain cells, which affects brain development and function.

Other Names for This Gene

- DYT17
- DYT18
- DYT9
- GLUT
- GLUT1
- GTR1_HUMAN

- MGC141895
- MGC141896
- solute carrier family 2 (facilitated glucose transporter), member 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLC2A1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6513[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SLC2A1%5BTIAB%5D%29+OR+%28%28GLUT%5BTIAB%5D%29+OR+%28GLUT1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER), MEMBER 1; SLC2A1 (https://omim.org/entry/138140)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6513)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SLC2A1[gene])

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Genomic Location

The *SLC2A1* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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