



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

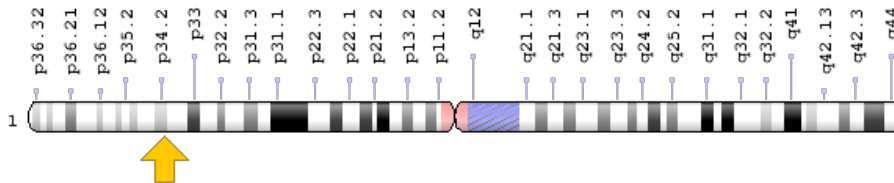
GLUT1 deficiency syndrome

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.

Chromosomal Location

Cytogenetic Location: 1p34.2, which is the short (p) arm of chromosome 1 at position 34.2

Molecular Location: base pairs 42,925,375 to 42,959,176 on chromosome 1 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DYT9
- DYT17
- DYT18
- GLUT
- GLUT1
- GTR1_HUMAN
- MGC141895
- MGC141896
- solute carrier family 2 (facilitated glucose transporter), member 1

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): GLUT1 Transports Glucose into Most Mammalian Cells
<https://www.ncbi.nlm.nih.gov/books/NBK21669/#A4044>

Clinical Information from GeneReviews

- Glucose Transporter Type 1 Deficiency Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1430>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?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%5BIa%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
<http://omim.org/entry/138140>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SLC2A1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC2A1%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:11005
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:6513>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6513>
- UniProt
<https://www.uniprot.org/uniprot/P11166>

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