SLC35A2 gene

solute carrier family 35 member A2

Normal Function

The *SLC35A2* gene provides instructions for making an enzyme called UDP-galactose translocator (UGT). This enzyme is involved in a process called glycosylation. During this process, complex chains of sugar molecules (oligosaccharides) are added to proteins and fats (lipids). Glycosylation modifies proteins and lipids so they can fully perform their functions. The UGT enzyme transfers a simple sugar called galactose to growing oligosaccharides at a particular step in the formation of the sugar chain. Once the correct number of sugar molecules are linked together, the oligosaccharide is attached to a protein or lipid.

Two versions of the enzyme, known as UGT1 and UGT2, are produced from the *SLC35A2* gene. These enzymes differ in only a few protein building blocks (amino acids) and can function together or separately in different areas of the cell.

Health Conditions Related to Genetic Changes

SLC35A2-congenital disorder of glycosylation

At least nine mutations in the *SLC35A2* gene have been found to cause *SLC35A2*-congenital disorder of glycosylation (*SLC35A2*-CDG). *SLC35A2*-CDG is an inherited condition that causes neurological problems (such as seizures, developmental delay, and intellectual disability) and abnormalities affecting other body systems.

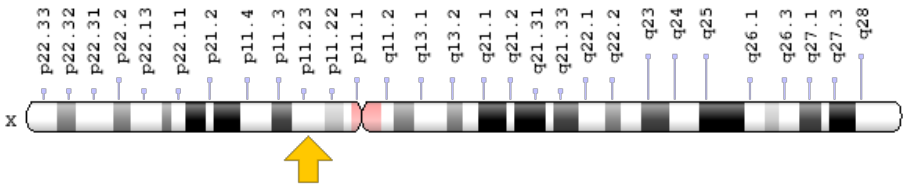
SLC35A2 gene mutations change single amino acids in the UGT enzyme or disrupt the way the gene's instructions are used to make the enzyme. These mutations can affect one or both versions of the enzyme and lead to the production of an abnormal enzyme with reduced or no activity. Without a properly functioning enzyme, glycosylation cannot proceed normally, and oligosaccharides are incomplete. The signs and symptoms of *SLC35A2*-CDG are likely due to impaired glycosylation of proteins and fats that are needed for the normal function of various organs and tissues.

In some individuals with *SLC35A2*-CDG, glycosylation becomes normal later in childhood. The cause of this apparent correction is unknown. The restoration of glycosylation in these individuals, however, does not seem to improve the signs and symptoms of *SLC35A2*-CDG.

Chromosomal Location

Cytogenetic Location: Xp11.23, which is the short (p) arm of the X chromosome at position 11.23

Molecular Location: base pairs 48,903,183 to 48,911,958 on the X chromosome (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- solute carrier family 35 (UDP-galactose transporter), member 2
- solute carrier family 35 (UDP-galactose transporter), member A2
- UDP-Gal-Tr
- UDP-galactose translocator
- UGALT
- UGAT
- UGT
- UGT1
- UGT2
- UGTL

Additional Information & Resources

Educational Resources

- Essentials of Glycobiology (second edition, 2009): Cellular Organization of Glycosylation
  https://www.ncbi.nlm.nih.gov/books/NBK1926/
- Molecular Biology of the Cell (fourth edition, 2002): What Is the Purpose of Glycosylation?
  https://www.ncbi.nlm.nih.gov/books/NBK26941/#A2354

Clinical Information from GeneReviews

- Congenital Disorders of N-Linked Glycosylation and Multiple Pathway Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1332

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC35A2%5BTIAB%5D%29+OR+%28%28UDP-Gal-Tr%5BTIAB%5D%29+OR+%28UDP-galactose+translocator%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

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 35 (UDP-GALACTOSE TRANSPORTER), MEMBER 2
  http://omim.org/entry/314375

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC35A2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:11022
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:7355
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7355
- Undiagnosed Diseases Network
  https://undiagnosed.hms.harvard.edu/genes/slc35a2/
- UniProt
  https://www.uniprot.org/uniprot/P78381

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