

## **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.

## 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

### Tests Listed in the Genetic Testing Registry

- Tests of SLC35A2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7355\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7355[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29>)

### Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 35 (UDP-GALACTOSE TRANSPORTER), MEMBER 2; SLC35A2 (<https://omim.org/entry/314375>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/7355>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SLC35A2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SLC35A2[gene]))

## References

- Dorre K, Olczak M, Wada Y, Sosicka P, Gruneberg M, Reunert J, Kurlemann G, Fiedler B, Biskup S, Hortnagel K, Rust S, Marquardt T. A new case of UDP-galactose

transporter deficiency (SLC35A2-CDG): molecular basis, clinical phenotype, and therapeutic approach. *J Inher Metab Dis*. 2015 Sep;38(5):931-40. doi: 10.1007/s10545-015-9828-6. Epub 2015 Mar 17. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/25778940>)

- Kimizu T, Takahashi Y, Oboshi T, Horino A, Koike T, Yoshitomi S, Mori T, Yamaguchi T, Ikeda H, Okamoto N, Nakashima M, Saitsu H, Kato M, Matsumoto N, Imai K. A case of early onset epileptic encephalopathy with de novo mutation in SLC35A2: Clinical features and treatment for epilepsy. *Brain Dev*. 2017 Mar;39(3):256-260. doi: 10.1016/j.braindev.2016.09.009. Epub 2016 Oct 12. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/27743886>)
- Kodera H, Nakamura K, Osaka H, Maegaki Y, Haginoya K, Mizumoto S, Kato M, Okamoto N, Iai M, Kondo Y, Nishiyama K, Tsurusaki Y, Nakashima M, Miyake N, Hayasaka K, Sugahara K, Yuasa I, Wada Y, Matsumoto N, Saitsu H. De novo mutations in SLC35A2 encoding a UDP-galactose transporter cause early-onset epileptic encephalopathy. *Hum Mutat*. 2013 Dec;34(12):1708-14. doi: 10.1002/humu.22446. Epub 2013 Oct 15. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24115232>)
- Ng BG, Buckingham KJ, Raymond K, Kircher M, Turner EH, He M, Smith JD, Eroshkin A, Szybowska M, Losfeld ME, Chong JX, Kozenko M, Li C, Patterson MC, Gilbert RD, Nickerson DA, Shendure J, Bamshad MJ; University of Washington Center for Mendelian Genomics; Freeze HH. Mosaicism of the UDP-galactose transporter SLC35A2 causes a congenital disorder of glycosylation. *Am J Hum Genet*. 2013 Apr;92(4):632-6. doi: 10.1016/j.ajhg.2013.03.012. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23561849>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3617373/>)

## Genomic Location

The *SLC35A2* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

**Last updated August 1, 2018**