

## SLC17A5 gene

solute carrier family 17 member 5

### Normal Function

The *SLC17A5* gene provides instructions for producing a protein called sialin that is located mainly on the membranes of lysosomes, compartments in the cell that digest and recycle materials. Sialin moves a molecule called free sialic acid, which is produced when certain proteins and fats are broken down, out of the lysosomes to other parts of the cell. Free sialic acid means that the sialic acid is not attached (bound) to other molecules.

Researchers believe that sialin may also have other functions in brain cells, in addition to those associated with the lysosomes, but these additional functions are not well understood.

### Health Conditions Related to Genetic Changes

#### Sialic acid storage disease

Approximately 20 mutations that cause sialic acid storage disease have been identified in the *SLC17A5* gene. Some of these mutations result in sialin that does not function normally; others prevent sialin from being produced. In a few cases, sialin is produced but not routed properly to the lysosomal membrane.

There are three forms of sialic acid storage disease. A particular *SLC17A5* mutation, found primarily in people from Finland and Sweden, causes the least severe form of this disorder known as Salla disease. This mutation replaces the protein building block (amino acid) arginine with the amino acid cysteine at position 39 of the sialin protein (written as Arg39Cys or R39C). Other *SLC17A5* gene mutations that have more damaging effects on sialin protein function cause the most severe form of the disorder, infantile free sialic acid storage disease. Individuals diagnosed with intermediate severe Salla disease have one copy of the *SLC17A5* gene with the Salla disease mutation in each cell, while the second copy of the gene bears a more severe mutation. The severity of signs and symptoms of intermediate severe Salla disease falls between those of Salla disease and infantile free sialic acid storage disease.

*SLC17A5* gene mutations that reduce or eliminate sialin activity result in a buildup of free sialic acid in the lysosomes. It is not known how this buildup, or disruption of other

possible functions of sialin in the brain, causes the specific signs and symptoms of sialic acid storage disease.

## Other Names for This Gene

- AST
- ISSD
- NSD
- S17A5\_HUMAN
- SD
- SIALIN
- SIASD
- SLD
- solute carrier family 17 (acidic sugar transporter), member 5
- solute carrier family 17, member 5

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of SLC17A5 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=26503\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=26503[geneid]))

## Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28sialic+acid+storage+disease%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

## Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 17 (ACIDIC SUGAR TRANSPORTER), MEMBER 5; SLC17A5 (<https://omim.org/entry/604322>)

## Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/26503>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SLC17A5\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SLC17A5[gene]))

## References

- Aula N, Kopra O, Jalanko A, Peltonen L. Sialin expression in the CNSimplicates extralysosomal function in neurons. *Neurobiol Dis.* 2004Mar;15(2):251-61. doi: 10.1016/j.nbd.2003.11.017. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15006695>)
- Eskelinen EL, Tanaka Y, Saftig P. At the acidic edge: emerging functions forlysosomal membrane proteins. *Trends Cell Biol.* 2003 Mar;13(3):137-45. doi:10.1016/s0962-8924(03)00005-9. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12628346>)
- Mach L. Biosynthesis of lysosomal proteinases in health and disease. *BiolChem.* 2002 May;383(5):751-6. doi: 10.1515/BC.2002.078. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12108539>)
- Morin P, Sagne C, Gasnier B. Functional characterization of wild-type andmutant human sialin. *EMBO J.* 2004 Nov 24;23(23):4560-70. doi:10.1038/sj.emboj.7600464. Epub 2004 Oct 28. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15510212>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC533050/>)
- Suwannarat P. Disorders of free sialic acid. *Mol Genet Metab.* 2005Jun;85(2):85-7. doi: 10.1016/j.ymgme.2005.04.005. No abstract available. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15973781>)

## Genomic Location

The *SLC17A5* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

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