SLC7A7 gene

solute carrier family 7 member 7

Normal Function

The *SLC7A7* gene provides instructions for producing a protein called y+L amino acid transporter 1 (y+LAT-1), which is involved in transporting certain protein building blocks (amino acids), namely lysine, arginine, and ornithine. The transportation of amino acids from the small intestine and kidneys to the rest of the body is necessary for the body to be able to make and use proteins. The y+LAT-1 protein forms one part (the light subunit) of a complex called the heterodimeric cationic amino acid transporter. This subunit is responsible for binding to the amino acids that are transported.

Health Conditions Related to Genetic Changes

Lysinuric protein intolerance

Variants (also called mutations) in the *SLC7A7* gene have been found to cause lysinuric protein intolerance. All of these variants impair the y+LAT-1 protein's ability to transport amino acids. People with lysinuric protein intolerance who are of Finnish descent typically have the same variant. This variant disrupts the way the gene's instructions are used to make the y+LAT-1 protein, causing the protein to be misplaced in the cell.

Variants in the y+LAT-1 protein disrupt the transportation of amino acids, reducing the amount of lysine, arginine, and ornithine in the body and increasing the amount of these amino acids in urine. The abnormal transportation and reduced amount of these amino acids in various tissues of the body leads to the signs and symptoms of lysinuric protein intolerance.

Other Names for This Gene

- LAT3
- LPI
- solute carrier family 7 (amino acid transporter light chain, y+L system), member 7
- y+LAT-1
- Y+LAT1
- YLAT1_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLC7A7 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9056[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SLC7A7%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 7 (CATIONIC AMINO ACID TRANSPORTER, y+ SYSTEM), MEMBER 7; SLC7A7 (https://omim.org/entry/603593)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/9056)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SLC7A7[gene])

References

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Genomic Location

The *SLC7A7* gene is found on chromosome 14 (https://medlineplus.gov/genetics/chromosome/14/).

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