



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 building blocks of protein (amino acids), namely lysine, arginine, and ornithine. The transportation of amino acids from the small intestines and kidneys to the rest of the body is necessary for the body to be able to 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

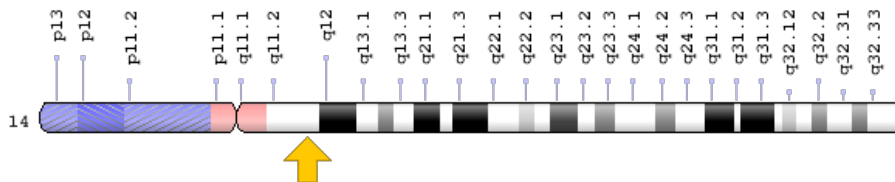
More than 40 mutations in the *SLC7A7* gene have been found to cause lysinuric protein intolerance. All of these mutations impair the y+LAT-1 protein's ability to transport amino acids. People with lysinuric protein intolerance who are of Finnish descent typically have the same mutation. This mutation (written as IVS6-2A>T) 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.

Mutations in the y+LAT-1 protein disrupt the transportation of amino acids, leading to a shortage of lysine, arginine, and ornithine in the body and an abnormally large amount of these amino acids in urine. The abnormal transportation and shortage of these amino acids in various tissues of the body leads to the signs and symptoms of lysinuric protein intolerance.

Chromosomal Location

Cytogenetic Location: 14q11.2, which is the long (q) arm of chromosome 14 at position 11.2

Molecular Location: base pairs 22,773,222 to 22,819,811 on chromosome 14 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Lysinuric protein intolerance is caused by defects in the transport of lysine, ornithine and arginine
<https://www.ncbi.nlm.nih.gov/books/NBK27982/#A3143>

Clinical Information from GeneReviews

- Lysinuric Protein Intolerance
<https://www.ncbi.nlm.nih.gov/books/NBK1361>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC7A7%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%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
<http://omim.org/entry/603593>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SLC7A7.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC7A7%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:11065
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:9056>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/9056>
- UniProt
<https://www.uniprot.org/uniprot/Q9UM01>

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