SLC6A19 gene
solute carrier family 6 member 19

Normal Function

The *SLC6A19* gene provides instructions for making a protein called system B(0) neutral amino acid transporter 1 (B₀AT1). This protein transports certain protein building blocks (amino acids), namely those with a neutral charge, into cells. B₀AT1 is found primarily in the membrane of intestinal cells that make up the brush border, which lines the walls of the intestine and absorbs nutrients from food. B₀AT1 transports the neutral amino acids from food into intestinal cells; from there the amino acids are released into the bloodstream to be used by the body. B₀AT1 is also found in the membrane of kidney cells, specifically in cells of the proximal tubules, which are structures that help to reabsorb nutrients and other materials into the blood and excrete unneeded substances into the urine. In the kidneys, B₀AT1 reabsorbs neutral amino acids into the bloodstream so they are not released in urine.

Health Conditions Related to Genetic Changes

Hartnup disease

At least 23 mutations in the *SLC6A19* gene have been found to cause Hartnup disease. This condition is characterized by increased levels of amino acids in the urine (aminoaciduria). Some individuals have episodes during which they exhibit skin rashes or movement or cognitive problems. Most of the mutations that cause Hartnup disease change single amino acids in the B₀AT1 protein, reducing its activity. A mutation that has been identified in multiple affected families replaces the amino acid aspartic acid with the amino acid asparagine at position 173 in the protein (written as Asp₁₇₃Asn or D₁₇₃N).

The reduced B₀AT1 activity leads to large amounts of neutral amino acids being removed from the body as waste. As a result, affected individuals are lacking (deficient) in certain amino acids and vitamins. Most affected people get the nutrients they need from a well-balanced diet. However, some individuals are nutrient-deficient due to their diet, illness, stress, or a variety of other reasons and can develop skin rashes or movement or psychiatric problems. Researchers believe that many of these features are related to a deficiency of vitamin B3 (niacin) and one of its main components, the amino acid tryptophan.
Chromosomal Location

Cytogenetic Location: 5p15.33, which is the short (p) arm of chromosome 5 at position 15.33

Molecular Location: base pairs 1,201,595 to 1,225,111 on chromosome 5 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

• B0AT1
• HND
• sodium-dependent amino acid transporter system B0
• sodium-dependent neutral amino acid transporter B(0)AT1
• solute carrier family 6 (neutral amino acid transporter), member 19
• system B(0) neutral amino acid transporter AT1
• system B0 neutral amino acid transporter

Additional Information & Resources

Educational Resources

• Biochemistry (fifth edition, 2002): Proteins Are Built from a Repertoire of 20 Amino Acids
  https://www.ncbi.nlm.nih.gov/books/NBK22379/

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC6A19%5BTIAB%5D%29+OR+%28B0AT1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Blan%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22
Catalog of Genes and Diseases from OMIM

• SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER), MEMBER 19
  http://omim.org/entry/608893

Research Resources

• Atlas of Genetics and Cytogenetics in Oncology and Haematology

• ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC6A19%5Bgene%5D

• HGNC Gene Symbol Report

• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:340024

• NCBI Gene

• UniProt
  https://www.uniprot.org/uniprot/Q695T7

Sources for This Summary


OMIM: SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER), MEMBER 19
http://omim.org/entry/608893

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