

## 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 (B<sup>0</sup>AT1). This protein transports certain protein building blocks (amino acids), namely those with a neutral charge, into cells. B<sup>0</sup>AT1 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. B<sup>0</sup>AT1 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. B<sup>0</sup>AT1 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, B<sup>0</sup>AT1 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 B<sup>0</sup>AT1 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 B<sup>0</sup>AT1 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.

## 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

### Tests Listed in the Genetic Testing Registry

- Tests of SLC6A19 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=340024\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=340024[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28SLC6A19%5BTIAB%5D%29+OR+%28B0AT1%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+3600+days%22%5Bdp%5D>)

### Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER), MEMBER 19; SLC6A19 (<https://omim.org/entry/608893>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/340024>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SLC6A19\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SLC6A19[gene]))

## References

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## Genomic Location

The *SLC6A19* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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