SLC19A3 gene

solute carrier family 19 member 3

Normal Function

The *SLC19A3* gene provides instructions for making a protein called a thiamine transporter, which moves a vitamin called thiamine into cells. Thiamine, also known as vitamin B1, is obtained from the diet. It is involved in many cellular processes, and is necessary for proper functioning of the nervous system. Molecules made from thiamine are important in the breakdown of sugars and protein building blocks (amino acids). Thiamine is also involved in the production of certain chemicals that relay signals in the nervous system (neurotransmitters).

Health Conditions Related to Genetic Changes

Biotin-thiamine-responsive basal ganglia disease

At least seven mutations in the *SLC19A3* gene have been identified in people with biotin-thiamine-responsive basal ganglia disease, a disorder that involves recurrent episodes of brain dysfunction (encephalopathy) and a variety of neurological problems that gradually get worse. *SLC19A3* gene mutations likely result in a protein with impaired ability to transport thiamine into cells, resulting in decreased absorption of the vitamin and leading to neurological dysfunction. Using medical imaging, abnormalities can be seen in several parts of the brain, including a group of structures called the basal ganglia, which help control movement, but the relationship between these specific brain abnormalities and the abnormal thiamine transporter is unknown.

Leigh syndrome

MedlinePlus Genetics provides information about Leigh syndrome

Other disorders

SLC19A3 gene mutations have also been identified in individuals with other neurological disorders whose signs and symptoms overlap those of biotin-thiamine-responsive basal ganglia disease (described above). These include a disorder called early infantile lethal encephalopathy and another disorder that begins in early infancy and causes seizures and brain deterioration (atrophy). A small number of individuals with signs and symptoms similar to those of the neurological disorders Leigh syndrome

and Wernicke encephalopathy have also been found to have *SLC19A3* gene mutations. It is unclear why mutations in this gene cause varying signs and symptoms in different individuals.

Other Names for This Gene

- BBGD
- S19A3_HUMAN
- solute carrier family 19 (thiamine transporter), member 3
- solute carrier family 19, member 3
- thiamine transporter 2
- THMD2
- thTr-2
- THTR2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLC19A3 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=80704[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SLC19A3%5BTIAB%5D%29+OR+%28%28thiamine+transporter+2%5BTIAB%5D%29+OR+%28BBGD%5BTIAB%5D%29+OR+%28THTR2%5BTIAB%5D%29+OR+%28thTr-2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3240+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 19 (THIAMINE TRANSPORTER), MEMBER 3; SLC19A3 (https://omim.org/entry/606152)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/80704)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SLC19A3[gene])

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Genomic Location

The *SLC19A3* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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