SLC25A19 gene

solute carrier family 25 member 19

Normal Function

The *SLC25A19* gene provides instructions for producing a protein that is a member of the solute carrier (SLC) family of proteins. Proteins in the SLC family transport various compounds across the membranes surrounding the cell and its component parts. The protein produced from the *SLC25A19* gene transports a molecule called thiamine pyrophosphate into the mitochondria, the energy-producing centers of cells. Thiamine pyrophosphate is involved in the functioning of a group of mitochondrial enzymes called the alpha-ketoglutarate dehydrogenase complex. This complex acts on a compound called alpha-ketoglutaric acid as part of an important series of reactions known as the citric acid cycle or Krebs cycle. The transport of thiamine pyrophosphate into the mitochondria is believed to be important in brain development.

Health Conditions Related to Genetic Changes

Amish lethal microcephaly

All known individuals with Amish lethal microcephaly have a mutation in which the protein building block (amino acid) alanine is substituted for the amino acid glycine at position 177 of the SLC25A19 protein, written as Gly177Ala or G177A. Researchers believe that this mutation interferes with the transport of thiamine pyrophosphate into the mitochondria and the activity of the alpha-ketoglutarate dehydrogenase complex, resulting in the abnormal brain development and the excess of alpha-ketoglutaric acid in the urine characteristic of Amish lethal microcephaly.

Leigh syndrome

MedlinePlus Genetics provides information about Leigh syndrome

Other Names for This Gene

- DNC
- DNC_HUMAN
- MCPHA
- mitochondrial deoxynucleotide carrier

- mitochondrial uncoupling protein 1
- MUP1
- solute carrier family 25 (mitochondrial deoxynucleotide carrier), member 19
- solute carrier family 25 (mitochondrial thiamine pyrophosphate carrier), member 19
- solute carrier family 25, member 19
- TPC

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLC25A19 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=60386[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SLC25A19%5BTIAB%5D%29+OR+%28%28DNC%5BTIAB%5D%29+OR+%28MUP1%5BTIAB%5D%29+OR+%28MCPHA%5BTIAB%5D%29+OR+%28mitochondrial+deoxynucleotide+carrier%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL THIAMINE PYROPHOSPHATE CARRIER), MEMBER 19; SLC25A19 (https://omim.org/entry/606521)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/60386)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SLC25A19[gene])

References

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Genomic Location

The *SLC25A19* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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