SLC46A1 gene

solute carrier family 46 member 1

Normal Function

The *SLC46A1* gene provides instructions for making a protein called the proton-coupled folate transporter (PCFT). PCFT is important for normal functioning of intestinal epithelial cells, which are cells that line the walls of the intestine. These cells have fingerlike projections called microvilli that absorb nutrients from food as it passes through the intestine. Based on their appearance, groups of these microvilli are known collectively as the brush border. PCFT is involved in the process of using energy to move certain B vitamins called folates across the brush border membrane for absorption, a mechanism called active transport. It is also involved in the transport of folates between the brain and the fluid that surrounds it (cerebrospinal fluid). Folates are important for many cell functions, including the production of DNA and its chemical cousin, RNA.

Health Conditions Related to Genetic Changes

Hereditary folate malabsorption

More than 10 mutations in the *SLC46A1* gene have been identified in people with hereditary folate malabsorption. These mutations cause the substitution of one protein building block (amino acid) for another amino acid in the PCFT protein, or result in a PCFT protein that is shorter than normal. The mutated PCFT protein has little or no activity. In some cases the abnormal protein is not transported to the cell membrane, and so it is unable to perform its function. PCFT inactivity impairs the body's ability to absorb folates from food, leading to the signs and symptoms of hereditary folate malabsorption.

Other Names for This Gene

- HCP1
- heme carrier protein 1
- MGC9564
- PCFT
- proton-coupled folate transporter
- solute carrier family 46 (folate transporter), member 1

- solute carrier family 46, member 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLC46A1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=113235[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SLC46A1%5BTIAB%5D%29+OR+%28%28proton-coupled+folate+transporter%5BTIAB%5D%29+OR+%28HCP1%5BTIAB%5D%29+OR+%28PCFT%5BTIAB%5D%29+OR+%28heme+carrier+protein+1%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 46 (FOLATE TRANSPORTER), MEMBER 1; SLC46A1 (https://omim.org/entry/611672)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/113235)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SLC46A1[gene])

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Genomic Location

The *SLC46A1* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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