SLC22A5 gene

solute carrier family 22 member 5

Normal Function

The *SLC22A5* gene provides instructions for making a protein called OCTN2 that is found in the heart, liver, muscles, kidneys, and other tissues. This protein is positioned within the cell membrane, where it transports a substance known as carnitine into the cell. Carnitine is mainly obtained from food and is needed to bring certain types of fats (fatty acids) into mitochondria, which are the energy-producing centers within cells. Fatty acids are a major source of energy for the heart and muscles. During periods without food (fasting), fatty acids become the most important energy source for the heart and other muscles.

Health Conditions Related to Genetic Changes

Primary carnitine deficiency

Many variants (also called mutations) in the *SLC22A5* gene have been found to cause primary carnitine deficiency. Some of these variants create a premature stop signal in the instructions for making the OCTN2 protein, resulting in an abnormally short, nonfunctional protein. Other variants change single protein building blocks (amino acids) in the OCTN2 protein or prevent the protein from being produced at all.

A lack of functional OCTN2 proteins create a shortage (deficiency) of carnitine within cells. Without carnitine, fatty acids cannot enter mitochondria and be used to make energy. Reduced energy production can lead to some of the features of primary carnitine deficiency, such as muscle weakness and hypoglycemia. Fatty acids can also build up in cells and damage the liver, heart, and muscles. This abnormal buildup causes the other signs and symptoms of the disorder.

Crohn's disease

MedlinePlus Genetics provides information about Crohn's disease

Other Names for This Gene

- CDSP
- high-affinity sodium dependent carnitine cotransporter

- novel organic cation transporter 2
- OCTN2
- organic cation transporter 5
- organic cation/carnitine transporter 2
- S22A5_HUMAN
- SCD
- solute carrier family 22 (organic cation transporter), member 5
- solute carrier family 22 (organic cation/carnitine transporter), member 5

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLC22A5 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6584[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SLC22A5%5BTIAB%5D%29+OR+%28solute+carrier+family+22+member+5%29+OR+%28OCTN2%5BTIAB%5D%29+AND+%28carnitine+deficiency%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 22 (ORGANIC CATION TRANSPORTER), MEMBER 5; SLC22A5 (https://omim.org/entry/603377)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6584)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SLC22A5[gene])

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Genomic Location

The *SLC22A5* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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