SLC25A20 gene

solute carrier family 25 member 20

Normal Function

The *SLC25A20* gene provides instructions for making a protein called carnitine-acylcarnitine translocase (CACT). This protein is essential for fatty acid oxidation, a multistep process that breaks down (metabolizes) fats and converts them into energy. Fatty acid oxidation takes place within mitochondria, which are the energy-producing centers in cells. A group of fats called long-chain fatty acids must be attached to a substance known as carnitine to enter mitochondria. Once these fatty acids are joined with carnitine, the CACT protein transports them into mitochondria. Carnitine is then removed from the long-chain fatty acid and transported back out of mitochondria by the CACT protein. Fatty acids are a major source of energy for the heart and muscles. During periods of fasting, fatty acids are also an important energy source for the liver and other tissues.

Health Conditions Related to Genetic Changes

Carnitine-acylcarnitine translocase deficiency

At least 27 mutations in the *SLC25A20* gene have been found to cause carnitine-acylcarnitine translocase (CACT) deficiency. Although these mutations change the structure of the CACT protein in different ways, they all lead to a shortage (deficiency) of the protein. Without enough functional CACT protein, long-chain fatty acids cannot be transported into mitochondria. As a result, these fatty acids are not converted to energy. Reduced energy production can lead to some of the features of CACT deficiency, such as low blood glucose (hypoglycemia) and low levels of the products of fat breakdown (hypoketosis). Fatty acids and long-chain acylcarnitines (fatty acids still attached to carnitine) may also build up in cells and damage the liver, heart, and muscles. This abnormal buildup causes the other signs and symptoms of the disorder.

Other Names for This Gene

- CAC
- CACT
- carnitine-acylcarnitine carrier
- carnitine/acylcarnitine translocase

- MCAT_HUMAN
- solute carrier family 25 (carnitine/acylcarnitine translocase), member 20

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLC25A20 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=788[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SLC25A20%5BTIAB%5D%29+OR+%28carnitine+translocase%5BTIAB%5D%29+OR+%28acylcarnitine+translocase%5BTIAB%5D%29+OR+%28carnitine/acylcarnitine+translocase%5BTIAB%5D%29%29+OR+%28CACT+gene%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 25 (CARNITINE/ACYLCARNITINE TRANSLOCASE), MEMBER 20; SLC25A20 (https://omim.org/entry/613698)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/788)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SLC25A20[gene])

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Genomic Location

The *SLC25A20* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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