CPT2 gene

carnitine palmitoyltransferase 2

Normal Function

The *CPT2* gene provides instructions for making an enzyme called carnitine palmitoyltransferase 2. This enzyme 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 inside mitochondria, carnitine palmitoyltransferase 2 removes the carnitine and adds a substance called coenzyme A. Long-chain fatty acids must be joined to coenzyme A before they can be metabolized to produce energy. 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 palmitoyltransferase II deficiency

More than 70 mutations in the *CPT2* gene have been found to cause carnitine palmitoyltransferase II (CPT II) deficiency. These mutations lead to reduced activity of carnitine palmitoyltransferase 2. Mutations that lead to extremely reduced enzyme activity typically cause the more severe forms of CPT II deficiency (a lethal neonatal form and a severe infantile hepatocardiomuscular form), while those that result in partially reduced enzyme activity usually lead to a less severe myopathic form of the disorder. The most common *CPT2* gene mutation replaces the protein building block (amino acid) serine with the amino acid leucine at position 113 (written as Ser113Leu or S113L) in the enzyme. This mutation accounts for about 60 percent of the mutations that cause the myopathic form of CPT II deficiency.

Without enough functioning carnitine palmitoyltransferase 2, long-chain fatty acids are not properly processed after they enter mitochondria and cannot be metabolized to produce energy. Reduced energy production can lead to some of the features of CPT II deficiency, such as muscle pain and weakness, 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

- CPT II
- CPT2_HUMAN
- CPTASE

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CPT2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1376[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CPT2%5BTIAB%5D%29+OR+%28carnitine+palmitoyltransferase+II%5BTIAB%5D%29%29+OR+%28%28CPT1%5BTIAB%5D%29+OR+%28CPTASE%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CARNITINE PALMITOYLTRANSFERASE II; CPT2 (https://omim.org/entry/600650)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1376)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CPT2[gene])

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Genomic Location

The *CPT2* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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