



CPT1A gene

carnitine palmitoyltransferase 1A

Normal Function

The *CPT1A* gene provides instructions for making an enzyme called carnitine palmitoyltransferase 1A, which is found in the liver. 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 cannot enter mitochondria unless they are attached to a substance known as carnitine. Carnitine palmitoyltransferase 1A connects carnitine to long-chain fatty acids so they can cross the inner membrane of mitochondria. Once these fatty acids are inside mitochondria, carnitine is removed and they can be metabolized to produce energy. During periods of fasting, long-chain fatty acids are an important energy source for the liver and other tissues.

Health Conditions Related to Genetic Changes

Carnitine palmitoyltransferase I deficiency

More than 20 mutations in the *CPT1A* gene have been found to cause carnitine palmitoyltransferase I (CPT I) deficiency. Most of these mutations change single protein building blocks (amino acids) within carnitine palmitoyltransferase 1A. Mutations in the *CPT1A* gene severely reduce or eliminate the activity of this enzyme. Without enough of this enzyme, carnitine is not attached to long-chain fatty acids. As a result, these fatty acids cannot enter mitochondria and be converted into energy. Reduced energy production can lead to some of the features of CPT I deficiency, such as low blood sugar (hypoglycemia) and low levels of the products of fat breakdown (hypoketosis). Fatty acids may also build up in cells and damage the liver, heart, and brain. This abnormal buildup causes the other signs and symptoms of the disorder.

Other disorders

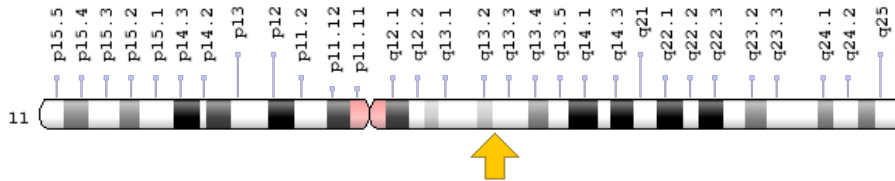
CPT1A gene mutations appear to increase the risk of a serious liver disorder that can develop in women during pregnancy. This disorder, called acute fatty liver of pregnancy, begins with abdominal pain and can rapidly progress to liver failure. Signs of acute fatty liver of pregnancy include an abnormal accumulation of fat in the liver, hypoglycemia, increased levels of ammonia in the blood (hyperammonemia), and abnormalities in liver enzymes. A small percentage of women who have a mutation in one copy of the *CPT1A* gene in each cell and are carrying a fetus with mutations in both copies of the *CPT1A* gene develop this maternal liver disease. Little is known

about the relationship between *CPT1A* gene mutations and liver problems in the mother during pregnancy.

Chromosomal Location

Cytogenetic Location: 11q13.3, which is the long (q) arm of chromosome 11 at position 13.3

Molecular Location: base pairs 68,754,620 to 68,844,410 on chromosome 11 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- carnitine palmitoyltransferase 1A (liver)
- carnitine palmitoyltransferase I, liver
- CPT1
- CPT1-L
- CPT1A_HUMAN
- L-CPT1

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Carnitine Carries Long-Chain Activated Fatty Acids into the Mitochondrial Matrix
<https://www.ncbi.nlm.nih.gov/books/NBK22581/#A3054>

Clinical Information from GeneReviews

- Carnitine Palmitoyltransferase 1A Deficiency
<https://www.ncbi.nlm.nih.gov/books/NBK1527>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CPT1A%5BTIAB%5D%29+OR+%28carnitine+palmitoyltransferase+1A%5BTIAB%5D%29%29+OR+%28%28carnitine+palmitoyltransferase+1A%5BTIAB%5D%29+OR+%28CPT1%5BTIAB%5D%29+OR+%28carnitine+palmitoyltransferase+I%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+1080+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- CARNITINE PALMITOYLTRANSFERASE I, LIVER
<http://omim.org/entry/600528>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CPT1A.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CPT1A%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2328
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:1374>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1374>
- UniProt
<https://www.uniprot.org/uniprot/P50416>

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