



## 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 the diet and is needed to bring certain types of fats (fatty acids) into mitochondria, 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 are also an important energy source for the liver and other tissues.

### Health Conditions Related to Genetic Changes

#### Primary carnitine deficiency

More than 60 mutations in the *SLC22A5* gene have been found to cause primary carnitine deficiency. Some of these mutations create a premature stop signal in the instructions for making the OCTN2 protein, resulting in an abnormally short, nonfunctional protein. Other mutations change single protein building blocks (amino acids) in the OCTN2 protein.

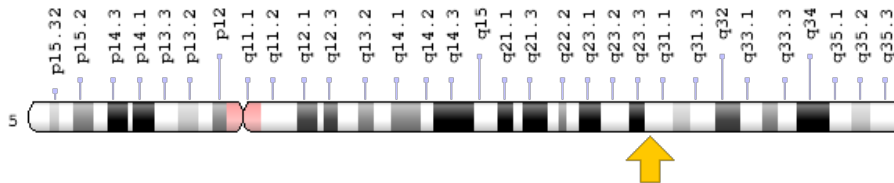
Mutations in the *SLC22A5* gene result in an absent or dysfunctional OCTN2 protein. As a result, there is a shortage (deficiency) of carnitine within cells. Without carnitine, fatty acids cannot enter mitochondria and be used to make energy. Reduced energy production can lead to some features of primary carnitine deficiency, such as muscle weakness and hypoglycemia. Fatty acids may also build up in cells and damage the heart, liver, and muscles. This abnormal buildup causes the other signs and symptoms of the disorder.

#### Crohn disease

## Chromosomal Location

Cytogenetic Location: 5q31.1, which is the long (q) arm of chromosome 5 at position 31.1

Molecular Location: base pairs 132,369,704 to 132,395,614 on chromosome 5 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

## 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

### 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

- Systemic Primary Carnitine Deficiency  
<https://www.ncbi.nlm.nih.gov/books/NBK84551>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?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  
<http://omim.org/entry/603377>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SLC22A5.html](http://atlasgeneticsoncology.org/Genes/GC_SLC22A5.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC22A5%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:10969](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:10969)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:6584>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6584>
- UniProt  
<https://www.uniprot.org/uniprot/O76082>
- University of Utah SLC22A5 Mutation Database  
[http://www.arup.utah.edu/database/OCTN2/OCTN2\\_welcome.php](http://www.arup.utah.edu/database/OCTN2/OCTN2_welcome.php)

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