

Primary carnitine deficiency

Description

Primary carnitine deficiency is a condition that prevents the body from using certain fats for energy, particularly during periods without food (fasting). Carnitine, a natural substance acquired mostly through food, is used by cells to process fats and produce energy.

Signs and symptoms of primary carnitine deficiency typically appear during infancy or early childhood and can include severe brain dysfunction (encephalopathy), a weakened and enlarged heart (cardiomyopathy), vomiting, muscle weakness, and low blood glucose (hypoglycemia). The severity of this condition varies among affected individuals. Some people with primary carnitine deficiency are asymptomatic, which means they do not have any signs or symptoms of the condition. All individuals with this disorder are at risk for sudden death.

Problems related to primary carnitine deficiency can be triggered by periods of fasting or by illnesses such as viral infections. This disorder is sometimes mistaken for Reye syndrome, a severe disorder that may develop in children who appear to be recovering from viral infections such as chickenpox or flu.

Frequency

Primary carnitine deficiency occurs in approximately 1 in 100,000 newborns worldwide. In Japan, this disorder affects 1 in every 40,000 newborns.

Causes

Variants (also called mutations) in the *SLC22A5* gene cause primary carnitine deficiency. This gene provides instructions for making a protein called OCTN2 that transports carnitine into cells. Cells need carnitine 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 of fasting, fatty acids become the most important energy source for the heart and other muscles.

Variants 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 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.

Learn more about the gene associated with Primary carnitine deficiency

- SLC22A5

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell must have a variant to cause the disorder. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Carnitine transporter deficiency
- Carnitine uptake defect
- Carnitine uptake deficiency
- CUD
- Renal carnitine transport defect
- Systemic carnitine deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Renal carnitine transport defect (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342788/>)

Genetic and Rare Diseases Information Center

- Systemic primary carnitine deficiency (<https://rarediseases.info.nih.gov/diseases/5104/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Primary carnitine deficiency%22](https://clinicaltrials.gov/search?cond=%22Primary%20carnitine%20deficiency%22))

Catalog of Genes and Diseases from OMIM

- CARNITINE DEFICIENCY, SYSTEMIC PRIMARY; CDSP (<https://omim.org/entry/212140>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28carnitine+uptake+defect%5BTIAB%5D%29+OR+%28carnitine+transporter+deficiency%5BTIAB%5D%29+OR+%28primary+carnitine+deficiency%5BTIAB%5D%29+OR+%28systemic+carnitine+deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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