What is carnitine palmitoyltransferase II deficiency?

Carnitine palmitoyltransferase II deficiency is an inherited disorder of long-chain fatty acid oxidation characterized by hypoketotic hypoglycemia, cardiomyopathy, seizures, muscle pain and weakness, and myoglobinuria. Individuals with carnitine palmitoyltransferase II deficiency have a defect in the production of the enzyme carnitine palmitoyltransferase 2, which plays an important role in fatty acid oxidation (the process that breaks down fats and converts them into energy). Signs and symptoms of carnitine palmitoyltransferase II deficiency are due to the buildup of fatty acids and long-chain acylcarnitines as well as reduced energy production in cells.

What are the symptoms of carnitine palmitoyltransferase II deficiency and what treatment is available?

Carnitine palmitoyltransferase II deficiency is a disease with variable severity and age of onset, even within families. There are three main forms based on severity and age at onset.

In the neonatal form symptoms are seen soon after birth. Life expectancy is a few days to a few months. Signs and symptoms may include:

- Hypoketotic hypoglycemia (low levels of ketones and blood sugar)
- Seizures
- Respiratory failure
- Liver failure
- Arrhythmia (irregular heartbeat)
- Cardiomyopathy (abnormal heart muscle)
- Abnormal brain and kidneys

In the infantile form symptoms are usually seen in the first year of life. Signs and symptoms may include:

- Metabolic episodes, often triggered by periods of fasting or illness, with hypoketotic hypoglycemia, seizures, hepatomegaly (enlarged liver), and arrhythmia
- Liver failure
- Nervous system damage
- Coma
- Sudden death

In the adult form symptoms are seen in childhood or adolescence. Signs and symptoms may include:

- Metabolic episodes triggered by exercise, stress, exposure to extreme temperatures, illness, general anesthesia, certain medications, or fasting
- Recurrent episodes of muscle pain and weakness
- Myoglobinuria (protein in the urine)
- Kidney failure

There is no cure for carnitine palmitoyltransferase II deficiency. The primary treatment is long-term dietary therapy, nutritional supplements, and avoidance of stressors such as fasting and exercise.

Carnitine palmitoyltransferase deficiency is included in newborn screening panels in most states in the United States.

How is carnitine palmitoyltransferase II deficiency inherited?

Carnitine palmitoyltransferase II deficiency is an autosomal recessive disease caused by mutations in the CPT2 gene. An individual who inherits one copy of a CPT2 gene mutation is a carrier and is not expected to have related

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Carnitine palmitoyltransferase II deficiency

health problems. An individual who inherits two CPT2 mutations, one from each parent, is expected to be affected with carnitine palmitoyltransferase II deficiency.

If both members of a couple are carriers of mutations in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for carnitine palmitoyltransferase II deficiency?
Carnitine palmitoyltransferase II deficiency is a rare condition that can occur in individuals of all races and ethnicities. Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?
If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?
A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?
Fatty Oxidation Disorders Family Support Group: [https://www.fodsupport.org/cpt2.htm](https://www.fodsupport.org/cpt2.htm)

References