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Carnitine palmitoyltransferase II deficiency

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.[1-4] 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)[1,2]. 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[2].

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[1,3]. There are three main forms based on severity and age at onset.[1-4]

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[3].

Carnitine palmitoyltransferase deficiency is included in newborn screening panels in most states in the United States[5].

How is carnitine palmitoyltransferase II deficiency inherited?

Carnitine palmitoyltransferase II deficiency is an autosomal recessive disease caused by mutations in the *CPT2* gene[1]. An individual who inherits one copy of a *CPT2* gene mutation is a carrier and is not expected to have related

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?

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/carnitine-palmitoyltransferase-ii-deficiency

Fatty Oxidation Disorders Family Support Group: https://www.fodsupport.org/cpt2.htm

References

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