

What is carnitine-acylcarnitine translocase deficiency?

Carnitine-acylcarnitine translocase deficiency is an inherited disorder of long-chain fatty acid oxidation characterized by neurological abnormalities, cardiomyopathy and arrhythmias, skeletal muscle damage, and liver dysfunction.¹ Individuals with carnitine-acylcarnitine translocase deficiency have a defect in the carnitine-acylcarnitine translocase enzyme, which is essential for fatty acid oxidation, the process that breaks down fats and converts them into energy.² Signs and symptoms of carnitine-acylcarnitine translocase deficiency are due to reduced energy production as well as the buildup of fatty acids and long-chain acylcarnitines in cells².

What are the symptoms of carnitine-acylcarnitine translocase deficiency and what treatment is available?

Carnitine-acylcarnitine translocase deficiency is a disease with variable severity and age at onset. Symptoms may include:^{1,2}

- Hypoketotic hypoglycemia (low levels of ketones and blood sugar)
- Hyperammonemia (excess ammonia in the blood)
- Respiratory problems
- Seizures
- Arrhythmia (irregular heartbeat)
- Hepatomegaly (enlarged liver)
- Cardiomyopathy (abnormal heart muscle)
- Lethargy (lack of energy)
- Coma
- Sudden death

Many infants with this condition do not survive the newborn period.² Some individuals have a less severe form of carnitine-acylcarnitine translocase deficiency and do not experience symptoms until early childhood.²

There is no cure for carnitine-acylcarnitine translocase deficiency. Treatment is supportive and generally includes avoidance of fasting as well as long-term dietary therapy with restriction of long-chain fatty acids¹.

Carnitine-acylcarnitine translocase deficiency is included in newborn screening panels in most states in the United States³.

How is carnitine-acylcarnitine translocase deficiency inherited?

Carnitine-acylcarnitine translocase deficiency is an autosomal recessive disease caused by mutations in the *SLC25A20* gene². An individual who inherits one copy of a *SLC25A20* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits *SLC25A20* two mutations, one from each parent, is expected to be affected with carnitine-acylcarnitine translocase 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-acylcarnitine translocase deficiency?

Carnitine-acylcarnitine translocase deficiency is a very rare condition that can occur in individuals of all races and ethnicities. Its worldwide prevalence is unknown.

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-acylcarnitine-translocase-deficiency>

The Screening, Technology And Research in Genetics (STAR-G):
<http://www.newbornscreening.info/Parents/fattyacid disorders/CAT.html>

References

1. Rubio-Gozalbo, M. E., Bakker, J. A., Waterham, H. R., Wanders, R. J. Carnitine-acylcarnitine translocase deficiency, clinical, biochemical and genetic aspects. *Mol. Aspects Med.* 25: 521-532, 2004.
2. Carnitine-acylcarnitine translocase deficiency. *Genetics Home Reference*. Available at: <http://ghr.nlm.nih.gov/condition/carnitine-acylcarnitine-translocase-deficiency>. Accessed on February 5, 2016
3. National Newborn Screening Status Report. November 2, 2014. Available at <http://genes-r-us.uthscsa.edu/sites/genes-r-us/files/nbsdisorders.pdf>. Accessed January 6, 2016.