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. 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.

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:

- 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. Some individuals have a less severe form of carnitine-acylcarnitine translocase deficiency and do not experience symptoms until early childhood.

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 acids.

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

How is carnitine-acylcarnitine translocase deficiency inherited?
Carnitine-acylcarnitine translocase deficiency is an autosomal recessive disease caused by mutations in the SLC25A20 gene. 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.
Carnitine-acylcarnitine translocase deficiency

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?
The Screening, Technology And Research in Genetics (STAR-G): [http://www.newbornscreening.info/Parents/fattyaciddisorders/CAT.html](http://www.newbornscreening.info/Parents/fattyaciddisorders/CAT.html)

References