

**Disease Name:** **CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY**  
(SOLUTE CARRIER FAMILY 25 (CARNITINE/ACYLCARNITINE TRANSLOCASE),  
MEMBER 20; CARNITINE-ACYLCARNITINE CARRIER; CACT DEFICIENCY)

**Classification:** Fatty acid oxidation defect

**Genetic  
Information:**

**Inheritance:** Autosomal recessive

**Population Incidence:** Rare, less than 30 confirmed cases

**Ethnic Incidence:** No known population at increased risk

**Gene & Location:** SLC25A20- on 3p21.31

**Common Mutation:** No known common mutations

**OMIM #** \*212138

**Disease  
Information:**

**Symptom Onset:** Two phenotypes, one with neonatal onset and the other with onset in infancy up to 3 years of age.

**Symptoms:** 80% of patients present in the first week of life with nonketotic hypoglycemia, hyperammonemia, hypotonia, liver dysfunction and cardiomyopathy. The majority of patients with neonatal onset have died of cardiopulmonary complications, progressive liver failure and/or hyperammonemia. Patients with a milder phenotype have developed hypoglycemia during infancy and early childhood, but not cardiomyopathy.

**Physical Findings:** No particular dysmorphisms.

**Treatment:** Aggressive treatment of hypoglycemia, hyperammonemia and prevention lipolysis in the neonate may be lifesaving. Diets low in long chain fats, high in carbohydrate with MCT supplement and frequent feeding have been helpful in several cases. Carnitine supplementation has been used, but efficacy is still not known.

**Natural History without treatment:** Overall mortality rate among reported patients is about 73%, most die by age 3 yrs. At least 20 sibling deaths (presumably from the same disorder) have been reported, so the neonatal lethality is likely very high. The more severe patients tend to have <1% residual enzyme activity, while the milder forms have 2-6% of residual activity.

**Natural History with treatment:** A few patients have been aggressively treated from diagnosis and are alive and well. The majority of patients have died, either in the neonatal period or up to 3 years, despite treatment. It is unknown whether prospective diagnosis and treatment will make a difference.

**Metabolic Information:** **Missing Enzyme & Location:** Carnitine-acylcarnitine translocase (CACT) is an inner mitochondrial membrane enzyme that mediates the transport long chain fatty acid acylcarnitines into the mitochondria and exports free carnitine to the cytosol. CACT is essential to fatty acid beta-oxidation and energy production in the mitochondria.

**MS/MS profile:** C0 (free carnitine)- low  
C16 (palmitoyl carnitine)- elevated  
C18:1 (linoleoyl carnitine)- elevated

**Prenatal testing:** Available via enzyme assay of amniocytes or CVS

**Miscellaneous Information:** Creatine kinase levels have also been elevated in at least 2 patients.

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