

Disease Name	
CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (CACT)	
<i>(SOLUTE CARRIER FAMILY 25 (CARNITINE/ACYLCARNITINE TRANSLOCASE), MEMBER 20; CARNITINE-ACYLCARNITINE CARRIER; CACT DEFICIENCY)</i>	
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:	Eighty percent 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 of 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 percent, most die by age three years. 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 <one percent residual enzyme activity, while the milder forms have two to six percent 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 three 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.

Credit:	<i>Prepared by the North West Regional Newborn Screening Program Judith Tuerck, RN, MS, and Lorinda Paradise at Oregon Health Services University in Portland, Oregon and by Sara Copeland MD, Iowa Neonatal Metabolic Screening Program.</i>	
Sites of Reference:	OMIM - Carnitine Acylcarnitine Translocase Deficiency www3.ncbi.nlm.nih.gov/htbin-post/Omim/dispim?212138	
Support Groups:	FOD Family Support Group www.fodsupport.org/ 805 Montrose Drive Greensboro, NC 24710 Contact: Deb Lee Gould Phone: (336) 547-8682 FODgroup@aol.com	