

Disease Name	
CARNITINE PALMITOYL TRANSFERASE I DEFICIENCY (CPT1)	
<i>(CARNITINE PALMITOYLTRANSFERASE IA DEFICIENCY; CPT I DEFICIENCY; CPT DEFICIENCY, HEPATIC, TYPE I)</i>	
Classification:	Fatty acid oxidation defect
Genetic Information	
Inheritance:	Autosomal recessive.
Population Incidence:	Rare, less than 50 patients identified.
Ethnic Incidence:	Increased in the Hutterite community and Inuit populations.
Gene & Location:	CPT IA- 11q13.1-13.5
Common Mutation:	P479L founder mutation in Inuit populations. G710E founder mutation in Hutterite community. Otherwise, all have been private mutations.
OMIM #	#255120; *600528
Disease Information	
Symptom Onset:	Usually presents between 8-18 months, but a few have presented in the first week of life.
Symptoms:	Initial symptoms have occurred with or after episodes of fasting, infection or diarrhea. Hypoketotic hypoglycemia, hyperammonemia, lethargy, vomiting and seizures proceed to coma and death if not recognized and treated aggressively. Hepatic dysfunction with elevated ammonia and enzymes is common. Creatine kinase elevation may be seen in acute episodes, although myoglobinuria is usually absent. No evidence of chronic muscle weakness has been documented. Renal tubular acidosis was seen in several patients. Transient hypertriglyceridemia was also seen. Recurrent episodes are common until about 5 years of age then seem to resolve. Several patients with CPT I have had, during acute episodes, cardiac involvement with poor contractility, cardiomegaly and arrhythmias, one with steatosis of the heart. These tend to resolve with treatment of the hypoglycemia. The mother of one affected infant had acute fatty liver of pregnancy during the last trimester.
Physical Findings:	Persistent neurologic deficit probably from the initial insult is common. Otherwise no dysmorphisms.
Treatment:	Avoid fasting, MCT oil supplement and avoid dietary long-chain fatty acids.
Natural History without treatment:	While most patients have survived infancy and acute hypoglycemic episodes, some have suffered permanent neurological deficits. Death has occurred during acute episodes in some cases.
Natural History with treatment:	Theoretically normal development if hypoglycemia and cardiac involvement can be prevented.
Metabolic Information	
Missing Enzyme & Location:	The enzyme CPT I converts long chain acyl-CoA substrates and carnitine to respective acylcarnitines for transport into the mitochondria.
MS/MS profile:	C0 (free carnitine) – elevated/normal. C16 with C18:1 – low. C0/C16 ratio – very elevated. C0/C18 ratio – very elevated.
Prenatal testing:	Possible to do enzyme analysis on amniocytes or CVS in patients with known risk.
Miscellaneous	Metabolite confirmation is difficult due to lack of characteristic acylcarnitine or

Information:	acylglycine markers- free and total carnitine levels will be normal to high. CPT I deficiency results in accumulation of toxic long chain substrates and impedes mitochondrial beta-oxidation. There is no correlation between enzyme activity and clinical severity.	
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:		
Support Groups:	FOD Family Support Group 805 Montrose Drive Greensboro, NC 24710 www.fodsupport.org/ Contact: Deb Lee Gould (336) 547-8682 FODgroup@aol.com	