

Disease Name	
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (CPT II)	
<i>(MYOPATHY WITH DEFICIENCY OF CARNITINE PALMITOYLTRANSFERASE II; CPT II DEFICIENCY, MYOPATHIC; HYPOGLYCEMIA, HYPOKETOTIC, WITH DEFICIENCY OF CARNITINE PALMITOYLTRANSFERASE II; CPT DEFICIENCY, HEPATIC, TYPE II)</i>	
Classification:	Fatty acid oxidations disorder
Genetic Information	
Inheritance:	Autosomal recessive.
Population Incidence:	Unknown.
Ethnic Incidence:	No known population at increased risk.
Gene & Location:	CPT2 gene- 1q32
Common Mutation:	Adult onset with common mutation-S113L in 60%; other forms without common mutations.
OMIM #	*600650; *600649; #255110
Disease Information	
Symptom Onset:	Three forms of disease: Lethal neonatal with onset during neonatal period; Infantile hepatocardiomyopathy form with onset in first year of life; and Myopathic form with onset in the first to sixth decade of life.
Symptoms:	<p>Three phenotypes of the disease:</p> <ol style="list-style-type: none"> 1. Lethal neonatal form: <ul style="list-style-type: none"> -Episodes of liver failure with hypoketotic hypoglycemia -Cardiomyopathy -Cardiac arrhythmias -Seizures and coma after fasting or infection -Facial abnormalities or structural malformations (e.g., cystic renal dysplasia, neuronal migration defects) 2. Infantile hepatocardiomyopathy form: <ul style="list-style-type: none"> -Liver failure -Cardiomyopathy -Seizures, hypoketotic hypoglycemia -Peripheral myopathy -Attacks of abdominal pain and headache 3. Myopathic form: <ul style="list-style-type: none"> -Myalgia and myoglobinuria after fasting and/or exercise or other physiological stress- heat, cold or illness -Muscle weakness during attacks -Asymptomatic between attacks
Physical Findings:	Minor facial dysmorphism and renal dysplasia in the neonatal form
Treatment:	Avoid fasting. Intravenous fluids and glucose when ill, hypoglycemic or with myoglobinuria. High carbohydrate, low fat diet. MCT oil supplement to supply the medium chains for beta-oxidation. Carnitine supplementation is controversial, as it may increase long-chain acylcarnitines and cause arrhythmia.
Natural History without treatment:	In the adult/ classic form recurrent myoglobinuria may lead to renal failure and death although most patients survive. Death or disability may occur during the infantile hypoketotic hypoglycemic episodes and death has been uniform in all neonatal presentations.

Natural History with treatment:	Goal is to avoid myoglobinuria and muscle pain in the myopathic form. Patients with the severe neonatal forms are not likely to benefit from treatment.	
Metabolic Information		
Missing Enzyme & Location:	CARNITINE PALMITOYLTRANSFERASE II catalyzes the last step of the carnitine-dependent entry of activated long-chain fatty acids into the mitochondria for beta-oxidation. The defect is in the conversion of the long-chain acylcarnitines to their corresponding acyl-CoA's upon transport into the mitochondria. The result is accumulation of long chain acylcarnitine in the mitochondrial matrix.	
MS/MS profile:	C0 (free carnitine)- low. C16 (palmitoyl carnitine)- elevated. C18:1 (linoleoyl carnitine)- elevated. C16/C2 or C18/C2- elevated.	
Prenatal testing:	DNA and enzyme analysis are available for at risk families.	
Miscellaneous Information:	May be some thermolability of the mutant enzyme in the adult form and explain the episodes triggered by acute febrile illness. Some studies have looked at protective effect of estrogen on enzyme function that may be effective in the milder forms.	
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 Palmitoyl Transferase Deficiency Type II www3.ncbi.nlm.nih.gov/htbin-post/Omim/disp/600650	
Support Groups:	<p>FOD Family Support Group 805 Montrose Drive Greensboro, NC 24710 www.fodsupport.org/ Contact: Deb Lee Gould (336) 547-8682 FODgroup@aol.com</p> <p>Association for Neuro-Metabolic Disorders (ANMD) PO Box 0202/L3220 1500 Medical Center Drive Ann Arbor, MI 48109-0202 313-763-4697 Fax: 313-764-7502</p>	<p>James William Lazzaro Foundation 4493 Liberty Road South Euclid, OH 44121 www.jwlsite.com/ Contact: Jamie Lazzaro (502) 254-2209 info@jwlsite.com</p> <p>Muscular Dystrophy Association (MDA) 3300 East Sunrise Drive Tucson, AZ 85718-3208 800-572-1717; 520-529-2000 mda@mdausa.org www.mdausa.org</p>