



Genetic Fact Sheets for Professionals

Fatty Acid Oxidation Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – [http:// www.newbornscreening.info](http://www.newbornscreening.info)

Disease name	Carnitine palmitoyl transferase deficiency, type 2
Alternate name(s)	CPTII
Acronym	CPT2, CPTII
Disease classification	Fatty Acid Oxidation Disorder
Variants	Yes
Variant name	Classical adult form
Symptom onset	Neonatal, infantile, adulthood
Symptoms	<p>Severe infantile form: hypoketotic hypoglycemia, severe cardiac involvement, renal malformations, arrhythmias, sudden death.</p> <p>Adult form: weakness, exercise intolerance, myoglobinuria.</p>
Natural history without treatment	Severe infantile form is often fatal.
Natural history with treatment	Even with treatment, the severe infantile form of CPT2 may be fatal.
Treatment	Avoidance of fasting, dietary supplementation with medium chain triglycerides (MCT), cornstarch supplementation, carnitine supplementation, intravenous glucose and carnitine in acute episodes.
Other	N/A
Physical phenotype	Renal malformations
Inheritance	Autosomal recessive

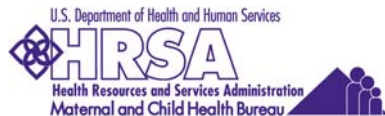
General population incidence	Rare
Ethnic differences	None
Population	N/A
Ethnic incidence	N/A
Enzyme location	Inner mitochondria membrane of skeletal muscle, cardiac and liver tissues
Enzyme function	Convert LCFA-carnitines to corresponding Acyl-CoA's
Missing enzyme	Carnitine palmitoyl transferase II
Metabolite changes	Low plasma and tissue carnitine levels, increased long-chain acylcarnitines, possible elevated CPK levels
Gene	CPT2
Gene location	1p32
DNA testing available	Yes
DNA testing detail	N/A
Prenatal testing	Enzyme analysis, analytes analysis and DNA analysis for known mutations.
MS/MS profile	C18:1, C18:2, C16, C16DC, C18:2DC, C18:1DC
OMIM link	www.ncbi.nlm.nih.gov/htbin-post/Omim/dispim?600650
Genetests link	www.genetests.org/servlet/access?prg=j&db=genetests&site=&id=12600&fcn=c&qry=3064&res=nous&res=nointl&key=DxfPGSWKadzr2&show_flag=c
Support group	FOD Family Support Group www.fodsupport.org Save Babies through Screening Foundation www.savebabies.org Genetic Alliance www.geneticalliance.org

Document Info

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