



## 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)

<b>Disease name</b>	<b>Carnitine palmitoyl transferase deficiency, type 1A (liver)</b>
<b>Alternate name(s)</b>	CPT1 deficiency, CPT1 liver
<b>Acronym</b>	CPT1, CPT1A
<b>Disease classification</b>	Fatty Acid Oxidation Disorder
<b>Variants</b>	Yes
<b>Variant name</b>	Carnitine palmitoyl transferase deficiency, type 1B - muscle type
<b>Symptoms</b>	Hypoketotic hypoglycemia, hepatomegaly, hyperammonemia, renal tubular acidosis.
<b>Natural history without treatment</b>	Possible developmental delay due to hypoglycemic episode, seizures, coma and death may occur.
<b>Natural history with treatment</b>	Attacks seem to get less frequent and less severe with age. Treatment cannot reverse developmental delay (if present).
<b>Treatment</b>	Avoidance of fasting, IV glucose during illnesses, dietary reduction of long chain fatty acids, dietary supplementation with medium chain fatty acids, cornstarch supplementation.
<b>Other</b>	Maternal complications in pregnancy including acute fatty liver of pregnancy and pre-eclampsia have been reported but may be coincidental.
<b>Physical phenotype</b>	None
<b>Inheritance</b>	Autosomal recessive
<b>General population incidence</b>	Rare
<b>Ethnic differences</b>	None
<b>Population</b>	N/A

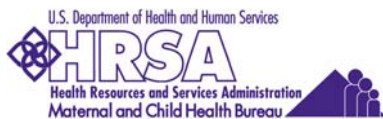
<b>Ethnic incidence</b>	N/A
<b>Enzyme location</b>	Liver, kidney, fibroblasts and heart
<b>Enzyme function</b>	Transfer long chain fatty acids into mitochondria
<b>Missing enzyme</b>	Carnitine palmitoyl transferase 1A
<b>Metabolite changes</b>	Elevated plasma carnitine level, absence of dicarboxylic aciduria, elevated CPK (possible).
<b>Gene</b>	CPT1
<b>Gene location</b>	11q13
<b>DNA testing available</b>	May be available on a research basis.
<b>DNA testing detail</b>	If a mutation in a proband is detected, DNA carrier screening is possible.
<b>Prenatal testing</b>	Protein analysis in CVS and amniocytes. If a mutation in a proband is detected, DNA prenatal diagnosis via CVS & amniocytes is possible.
<b>MS/MS profile</b>	Absence of long and medium chain acyl carnitines
<b>OMIM link</b>	<a href="http://www.ncbi.nlm.nih.gov/htbin-post/Omim/dispmim?255120">www.ncbi.nlm.nih.gov/htbin-post/Omim/dispmim?255120</a>
<b>Genetests link</b>	<a href="http://www.genetests.org/servlet/access?prg=j&amp;db=genestar&amp;site=&amp;fcn=d&amp;id=12600&amp;qry=68896&amp;res=nous&amp;res=nointl&amp;key=BtztHtFOOy6RR&amp;show_flag=c">www.genetests.org/servlet/access?prg=j&amp;db=genestar&amp;site=&amp;fcn=d&amp;id=12600&amp;qry=68896&amp;res=nous&amp;res=nointl&amp;key=BtztHtFOOy6RR&amp;show_flag=c</a>
<b>Support group</b>	FOD Family Support Group <a href="http://www.fodsupport.org">www.fodsupport.org</a>  Save Babies through Screening Foundation <a href="http://www.savebabies.org">www.savebabies.org</a>  Genetic Alliance <a href="http://www.geneticalliance.org">www.geneticalliance.org</a>

## Document Info

<b>Created by</b>	www.newbornscreening.info
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