



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	Medium-chain acyl-CoA dehydrogenase deficiency
Alternate name(s)	None
Acronym	MCADD
Disease classification	Fatty Acid Oxidation Disorder
Variants	N/A
Variant name	N/A
Symptom onset	Typically 6-24 months but ranges from neonatal to adult
Symptoms	Recurrent episodes of hypoglycemia, vomiting, coma, sudden death and possible seizures. Hepatomegaly usually present.
Natural history without treatment	Metabolic episodes can cause developmental and physical delays, neurologic impairment and sudden death.
Natural history with treatment	Normal intellect and physical functioning expected.
Treatment	Dietary: avoid fasting, low-fat diet (<30% of dietary fat), carnitine supplementation, cornstarch supplementation.
Other	N/A
Physical phenotype	None
Inheritance	Autosomal recessive

General population incidence	1/15,000
Ethnic differences	Yes
Population	Incidence higher in Northern Europeans and U.S Caucasians.
Ethnic incidence	Approximately 1/70 carrier rate
Enzyme location	Liver, heart, muscle and fibroblasts
Enzyme function	Mitochondrial beta-oxidation of fat stores
Missing enzyme	Medium-chain acyl-CoA dehydrogenase
Metabolite changes	Increased medium chain fatty acids, increased glycine/carnitine esters, increased dicarboxylic acids.
Gene	ACADM
Gene location	1p31
Gene description	12 exons coding for a 44kb gene
DNA testing available	Yes
DNA testing detail	One mutation K304E (985A) accounts for majority of disease incidence, 80% of affected individuals are homozygous for this mutation and 18% of affected individuals are heterozygous.
Prenatal testing	DNA and enzymatic testing
Carrier testing	May be possible by DNA depending on proband's genotype
MS/MS profile	Elevated C10:1, C8, C6
Genetests link	www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=74IDAJIXrlu&gry=&fcn=y&fw=bjWo&filename=/profiles/mcad/index.html

OMIM link www.ncbi.nlm.nih.gov/htbin-post/Omim/dispim?201450

Support group FOD Family Support Group
www.fodsupport.org

Organic Acidemia Association
www.oaanews.org

Save Babies through Screening Foundation
www.savebabies.org

Genetic Alliance
www.geneticalliance.org

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