



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	Very long-chain acyl-CoA dehydrogenase deficiency
Alternate name(s)	N/A
Acronym	VLCADD
Disease classification	Fatty Acid Oxidation Disorder
Variants	Yes
Variant name	With and without cardiomyopathy
Symptom onset	Primarily neonatal but some variability.
Symptoms	Hypoketotic hypoglycemia, hepatomegaly, myopathy, cardiomyopathy, adult-onset myopathy.
Natural history without treatment	Sudden infant death due to cardiac abnormalities is common.
Natural history with treatment	Diagnosis and treatment seem to decrease risk for sudden death.
Treatment	Avoidance of fasting, high carbohydrate, low-fat diet supplemented with MCT oil, IV glucose during illness, cornstarch supplementation, avoidance of long chain fatty acids, possible carnitine supplementation.
Other	May have history of a sibling dying of SIDS.
Physical phenotype	No particular dysmorphisms. Cardiomyopathy in infants.
Inheritance	Autosomal recessive

General population incidence	Rare – exact incidence not known
Ethnic differences	None reported
Population	N/A
Ethnic incidence	N/A
Enzyme location	Mitochondrial matrix, heart, liver
Enzyme function	Long chain fatty acid beta-oxidation
Missing enzyme	Very long-chain acyl-CoA dehydrogenase
Metabolite changes	Dicarboxylic aciduria, decreased urinary carnitine at times of illness, plasma free carnitine - normal to low, increased plasma long-chain acylcarnitines mildly increased ammonia, lactate and creatine kinase.
Gene	ACADVL
Gene location	17p11.2-p11.1
DNA testing available	Available on a research basis
DNA testing detail	No common mutations have been found. If a mutation in a proband is detected, DNA carrier screening is possible.
Prenatal testing	Enzyme and protein analysis. If a mutation in a proband is detected, DNA prenatal diagnosis via CVS or amniocytes is possible.
MS/MS profile	Elevated C16:1, C14:2, C14:1, C18:1
OMIM link	www.ncbi.nlm.nih.gov/htbin-post/Omim/dispim?201475
Genetests link	www.genetests.org/servlet/access?prg=j&db=genetests&site=&id=12600&fcn=c&qry=3179&res=nous&res=nointl&key=qaGe-CpasReRZ&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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