



## 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>Short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency</b>
<b>Alternate name(s)</b>	L-3-@hydroxyacyl-CoA dehydrogenase deficiency, SCHAD deficiency
<b>Acronym</b>	SCHAD or SCHADD
<b>Disease classification</b>	Fatty Acid Oxidation Disorder
<b>Inheritance</b>	Autosomal recessive
<b>Variants</b>	None known
<b>Variant name</b>	N/A
<b>Symptom onset</b>	Extremely variable – neonatal onset to adult onset. In addition, SCHADD has been implicated as a cause in a small minority of SIDS cases.
<b>Symptoms</b>	Hypoketotic hypoglycemia, recurrent myoglobinuria, encephalopathy, cardiomyopathy, ketotic hypoglycemia, failure to thrive, hypotonia and liver problems.
<b>Natural history without treatment</b>	Extremely variable. Neonatal presentation consisted of difficulty with feeding, failure to thrive, hypotonia and an elevated CPK. A sixteen year old presented with recurrent myoglobinuria and hypoketotic hypoglycemia.
<b>Natural history with treatment</b>	Treatment should prevent liver problems. The effectiveness of treatment in managing other symptoms is not known. Treatment will not reverse CNS damage due to hypoglycemic episodes.
<b>Treatment</b>	Avoidance of fasting, low-fat diet, carnitine supplementation, possible cornstarch supplementation.
<b>Other</b>	N/A
<b>Physical phenotype</b>	None reported

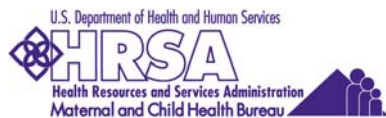
<b>Inheritance</b>	Autosomal recessive
<b>General population incidence</b>	Rare, exact incidence unknown
<b>Ethnic differences</b>	None known
<b>Population</b>	N/A
<b>Ethnic incidence</b>	N/A
<b>Enzyme location</b>	Mitochondria of liver, kidney, heart and muscle tissue
<b>Enzyme function</b>	Catalyzes the reversible dehydrogenation of 3-hydroxyacyl-CoAs to their corresponding 3-ketoacyl-CoAs
<b>Missing enzyme</b>	Short-chain 3-hydroxyacyl-CoA dehydrogenase
<b>Metabolite changes</b>	Increased adipic, suberic, sebacic and 3-hydroxydicarboxylic acids, and increased carnitine esters in blood. Elevated insulin and elevated creatinine phosphokinase. However, this was based on a small number of patients.
<b>Gene</b>	SCHAD or HADHSC gene
<b>Gene location</b>	4q22-q26
<b>DNA testing available</b>	Research only
<b>DNA testing detail</b>	N/A
<b>Prenatal testing</b>	Analyte assay on amniocentesis
<b>MS/MS Profile</b>	N/A
<b>OMIM link</b>	<a href="http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=601609">www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=601609</a>
<b>Genetests link</b>	<a href="http://www.genetests.org/servlet/access?prg=j&amp;db=genetests&amp;site=gt&amp;id=8888891&amp;fcn=c&amp;qry=26811&amp;res=nous&amp;res=nointl&amp;key=XFQ7rdtrg2ZHx&amp;show_flag=c">www.genetests.org/servlet/access?prg=j&amp;db=genetests&amp;site=gt&amp;id=8888891&amp;fcn=c&amp;qry=26811&amp;res=nous&amp;res=nointl&amp;key=XFQ7rdtrg2ZHx&amp;show_flag=c</a>
<b>Support group</b>	Fatty Oxidation Disorder (FOD) Family Support Group <a href="http://www.fodsupport.org">www.fodsupport.org</a>  Organic Acidemia Association <a href="http://www.oaanews.org">www.oaanews.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

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