



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	Short-chain acyl-CoA dehydrogenase deficiency
Alternate name(s)	N/A
Acronym	SCADD
Disease classification	Fatty Acid Oxidation Disorder
Variants	Yes
Variant name	Late-onset with chronic myopathy
Symptom onset	Neonatal - but very variable; may be asymptomatic.
Symptoms	Neonatal - failure to thrive, hypotonia, metabolic acidosis, seizures and developmental delay.
Natural history without treatment	Developmental delay, hypotonia and muscle weakness have been observed, but the vast majority of patients detected via MS/MS newborn screening have been entirely asymptomatic.
Natural history with treatment	The efficacy of treatment is unknown.
Treatment	Carnitine supplementation, restriction of dietary fat. A few patients have shown improvements on riboflavin supplements.
Other	Acute fatty liver of pregnancy and HELLP syndrome have been reported as maternal complications in pregnancy, but may be coincidental.
Physical phenotype	None reported
Inheritance	Autosomal recessive
General population incidence	1:40,000 – 1:100,000
Ethnic differences	None

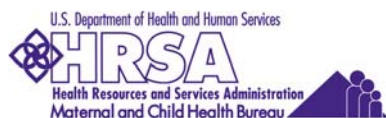
Population	N/A
Ethnic incidence	N/A
Enzyme location	Mitochondrial matrix in liver, muscle, fibroblasts
Missing enzyme	Short-chain acyl-CoA dehydrogenase
Metabolite changes	Ethylmalonic acid in urine, methylsuccinate and butyrylglycines in urine.
Gene	ACADS
Gene location	12q22-qter
Gene description	10 exons coding for a 13kb gene. No common disease mutations have been identified. Two common SCAD "disease susceptibility" mutations (625G>) (511C>T) are present in 14% of the general population. 69% of persons with ethylmalonic aciduria are either hetero/homozygous for these susceptibility mutations.
DNA testing available	Yes - but interpretation of results is difficult.
DNA testing detail	Limited mutational hotspots and common susceptibility alleles
Prenatal testing	Enzymatic
MS/MS profile	Elevated C4 Cbutyrylcarnitine
OMIM link	www.ncbi.nlm.nih.gov/htbin-post/Omim/dispim?606885
Genetests link	www.genetests.org/servlet/access?prg=j&db=genestar&site=&fcn=d&id=12600&qry=3176&res=nous&res=nointl&key=BtZrHtFOOy6RR&show_flag=c
Support groups	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

Document Info

Created by	www.newbornscreening.info
Reviewed by	HI, CA, OR and WA metabolic specialists
Review date	May 2, 2005
Updated on	N/A

DISCLAIMER:

THIS INFORMATION DOES NOT PROVIDE MEDICAL ADVICE. All content ("Content"), including text, graphics, images and information are for general informational purposes only. You are encouraged to confer with your doctor or other health care professional with regard to information contained on this information sheet. After reading this information sheet, you are encouraged to review the information carefully with your doctor or other healthcare provider. The Content is not intended to be a substitute for professional medical advice, diagnosis or treatment. NEVER DISREGARD PROFESSIONAL MEDICAL ADVICE, OR DELAY IN SEEKING IT, BECAUSE OF SOMETHING YOU HAVE READ ON THIS INFORMATION SHEET.



This project is supported by a grant from the Maternal and Child Health Bureau, Health Resources and Service Administration, Genetic Services Branch, MCH Project #:1H46 MC 00189-03 <http://mchb.hrsa.gov>