Disease Name	
SHORT-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (SCADD)	
(ACADS DEFICIENCY; SCADH DEFICIENCY; SCAD DEFICIENCY)	
Classification:	Fatty acid oxidation defect
Genetic Information	
Inheritance:	Autosomal Recessive.
Population Incidence:	1:40,000-1:100,000
Ethnic Incidence:	No known population at increased risk.
Gene & Location:	ACADS, SCAD- 12q22-qter
Common Mutation:	No common disease mutations have been identified. Two common SCAD "disease susceptibility" mutations (625G>A) (511C>T) are present in 14 percent of the general population. 69 percent of persons with ethylmalonic aciduria are either heterozygous or homozygous for these susceptibility mutations.
OMIM #	*606885; #201470
Disease Information	
Symptom Onset:	Only 20 SCAD deficient patients have been described. The majority became symptomatic in the first week to three months of life, four before 24 months and two presented as adults.
Symptoms:	About 50 percent of patients have hypotonia and developmental delay. Variable symptoms are present in patients presenting early, including seizures, respiratory distress, acidosis, poor feeding, vomiting or failure to thrive, and one death. Only one patient had hypoglycemia. Acute symptoms in most patients did not recur. Both adults presented with proximal muscle weakness associated with periods of pain, nausea/vomiting and shortness of breath.
Physical Findings:	No specific dysmorphology.
Treatment:	Supportive standard treatment of symptoms. Common fatty acid oxidation therapies including low fat diet, carnitine and riboflavin supplements have not improved hypotonia or developmental delay. One patient with cyclic vomiting improved on frequent feeding.
Natural History without treatment:	Highly variable, from death in the neonatal period to probable asymptomatic persons. It is reassuring that some of the early onset patients completely recovered and have had normal growth and development into adulthood. Long-term outcome beyond adolescence is unknown, specifically the development of further muscle weakness. Environmental stressors are suspected to precipitate symptoms in some cases, but they are not well understood.
Natural History with treatment:	There is no proven efficacy of the usual treatments for fatty acid oxidation disorders, with the possible exception of avoiding fasting.
Metabolic Information	
Missing Enzyme & Location:	Short chain acyl-CoA dehydrogenase, liver, muscle and fibroblasts. SCAD is one of three enzymes that catalyze the last step in mitochondrial beta-oxidation of fatty acids.
MS/MS profile:	C4 (butyryl/ isobutyryl carnitine)- elevated. C4/C2- elevated. C4/C3- elevated.
Prenatal testing:	If both gene mutations are known theoretically possible.
Miscellaneous Information:	Ethylmalonic acid and methylsuccinate are elevated in the urine. Maternal pregnancy complications include HELLP or acute fatty liver of pregnancy (AFLP) with some affected SCAD fetuses.

	Some patients will have isolated excretion of C4 and ethylmalonic acid	
Credit:	*Prepared by the North West Regional Newborn Screening Program Judith Tuerck, RN, MS, and Lorinda Paradise at Oregon Health Services University in Portland, Oregon and by Sara Copeland MD, Iowa Neonatal Metabolic Screening Program.*	
Sites of Reference:	**NORD - Short Chain Acyl CoA Dehydrogenase Deficiency (SCAD)** www.rarediseases.org/search/rdbdetail_abstract.html?disname=Short%20Chain%20Acyl%20CoA%20Dehydrogenase%20Deficiency%20%28SCAD%29 **Save Babies Through Screening Foundation, Inc** Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD) http://www.savebabies.org/diseasedescriptions/scad.php **OMIM - Short-Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)** www.ncbi.nlm.nih.gov/htbin-post/Omim/dispmim?201470	
Support Groups:	**FOD Family Support Group** 805 Montrose Drive Greensboro, NC 24710 www.fodsupport.org/ Contact: Deb Lee Gould (336) 547-8682 FODgroup@aol.com **United Mitochondrial Disease Foundation** P.O. Box 1151 Monroeville, PA 15146-1151 www.umdf.org/ (412) 793-8077 info@umdf.org	**James William Lazzaro Foundation** 4493 Liberty Road South Euclid, OH 44121 www.jwlsite.com/ Contact: Jamie Lazzaro (502) 254-2209 info@jwlsite.com **Children Living with Inherited Metabolic Diseases (CLIMB)** Climb Building 176 Nantwich Road Crewe, CW2 6BG United Kingdom (+44) 0870 7700 326 Fax: (+44) 0870 7700 327 steve@climb.org.uk www.climb.org.uk