

Disease Name	
VERY LONG-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (VLCADD)	
<i>(VLCAD DEFICIENCY)</i>	
Classification:	Fatty acid oxidation disorder
Genetic Information	
Inheritance:	Autosomal recessive.
Population Incidence:	Unknown.
Ethnic Incidence:	No known population at increased risk.
Gene & Location:	ACADVL, VLCAD- 17p11.2-p11.1
Common Mutation:	No common mutations seen.
OMIM #	*201475
Disease Information	
Symptom Onset:	Variable, depending on the phenotype, ranging from neonatal to adult onset.
Symptoms:	<p>Approximately 50 percent present as infants with nonketotic hypoglycemia, hepatic dysfunction and cardiomyopathy, and this has been generally lethal.</p> <p>Thirty-three percent present in late infancy or childhood with episodes of nonketotic hypoglycemia and hepatic dysfunction but no cardiac involvement. There is generally a mildly increased ammonia, lactate, and creatine kinase.</p> <p>Approximately 20 percent present as adolescents or adults with symptoms limited to muscle fatigue, rhabdomyolysis and myoglobinuria triggered by exercise or fasting. There is no hypoglycemia or cardiac involvement.</p>
Physical Findings:	No particular dysmorphisms. Cardiomyopathy in infants.
Treatment:	The mainstay of treatment is a high carbohydrate, low fat diet supplemented with MCT oil and strict avoidance of fasting and prolonged exercise. Aggressive support with calories and fluid is needed for intercurrent illnesses. Carnitine use is controversial.
Natural History without treatment:	Patients with the infantile form of the disease usually die in the first year of life. The late infantile hepatic presentation children will die without treatment. The adult form can progress to renal failure if the myoglobinuria is not addressed.
Natural History with treatment:	The infantile form is generally fatal, although there are now reports of survivors and complete resolution of cardiomyopathy with early diagnosis and treatment. The later onset patients can survive if treated appropriately. In general the outcome is believed to be good for patients who are identified presymptomatically.
Metabolic Information	
Missing Enzyme & Location:	Defect in palmitoyl-CoA dehydrogenase. Responsible for reducing acyl-CoA's of chain lengths C14-C20. This is the first and rate-limiting step in the beta-oxidation of fatty acids by the mitochondria for energy metabolism.
MS/MS profile:	C14:1 (tetradecenoyl carnitine)- elevated. C14:1/C12:1 ratio – elevated.
Prenatal testing:	Prenatal diagnosis is possible in families with a previously affected child.
Miscellaneous Information:	In the mouse model, there have been arrhythmias and death even in older mice. Confirmatory and diagnostic metabolic testing may be normal even in patients with a known VLCAD mutation.

Credit:	<i>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.</i>	
Sites of Reference:	<p>OMIM - Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD) www.ncbi.nlm.nih.gov/htbin-post/Omim/dispim?201475</p> <p>Save Babies Through Screening Foundation, Inc Very Long Chain Acyl-CoA Dehydrogenase Deficiency http://www.savebabies.org/diseasesdescriptions/vlcad.php</p>	
Support Groups:	<p>FOD Family Support Group 805 Montrose Drive Greensboro, NC 24 www.fodsupport.org/710 Contact: Deb Lee Gould (336) 547-8682 FODgroup@aol.com</p> <p>United Mitochondrial Disease Foundation P.O. Box 1151 Monroeville, PA 15146-1151 www.umdf.org/ (412) 793-8077 info@umdf.org</p>	<p>James William Lazzaro Foundation 4493 Liberty Road South Euclid, OH 44121 www.jwlsite.com/ Contact: Jamie Lazzaro (502) 254-2209 info@jwlsite.com</p>