

<b>Disease Name</b>	
<b>MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (MCADD)</b>	
<i>(ACADM DEFICIENCY; MCAD DEFICIENCY; MCADD DEFICIENCY)</i>	
<b>Classification:</b>	Fatty acid oxidation defect
<b>Genetic Information</b>	
<b>Inheritance:</b>	Autosomal recessive.
<b>Population Incidence:</b>	1/12,000
<b>Ethnic Incidence:</b>	Approximately 1/70 carrier rate; incidence higher in Northern Europeans and North American Caucasians.
<b>Gene &amp; Location:</b>	ACADM, 1p31
<b>Common Mutation:</b>	80% carry A985G; T199C is 2nd most common mutation.
<b>OMIM #</b>	#201450; *607008
<b>Disease Information</b>	
<b>Symptom Onset:</b>	Typically 3-24 months, variable even within family member with identical gene mutations and can range from neonatal to adult. MCADD may account for about three percent of SIDS deaths.
<b>Symptoms:</b>	Episodes of hypoketotic hypoglycemia, encephalopathy, coma or SIDS triggered by a common illness and/or a period of fasting. In 18 percent of cases, sudden death is the initial presenting manifestation and up to 25 percent of patients die during their first episode. Survivors may have significant developmental disability, muscle weakness, failure to thrive and cerebral palsy.
<b>Physical Findings:</b>	No particular physical findings or dysmorphisms.
<b>Treatment:</b>	Treatment is simple and effective: avoid fasting in patients of all ages; feeding increments depend on age of patient. It is imperative that glucose supplementation and hydration be provided during times of illness. A tangible clinical benefit of carnitine supplementation in patients with MCADD deficiency has not been proven, but several authors recommend oral supplementation with 50mg/kg/day to correct the observed secondary carnitine deficiency and to enhance the elimination of toxic metabolites.
<b>Natural History without treatment:</b>	Mortality rate of up to 25 percent when untreated. Metabolic episodes can cause developmental and physical delays from hypoglycemia and liver insult.
<b>Natural History with treatment:</b>	Most physicians familiar with MCADD presume that patients will develop normally if diagnosed early and treated in order to avoid hypoglycemia and liver dysfunction. However, this has not been proven. Death on the other hand, has not been reported once the diagnosis is made if treatment is carried out. Of survivors, diagnosed by clinical symptoms, 37 percent have neurodevelopment problems.
<b>Metabolic Information</b>	
<b>Missing Enzyme &amp; Location:</b>	Medium chain acyl-CoA dehydrogenase is found in the liver, heart, muscle and fibroblasts. Located in mitochondria and involved in fatty acid beta-oxidation. These patients are unable to metabolize C6-C10 length fatty acids.
<b>MS/MS profile:</b>	C6 (hexanoyl carnitine)- elevated. C8 (octanoyl carnitine)- elevated. C10 (decanoyl carnitine)- elevated. C10:1 (decenoyl carnitine)- elevated.

	C8/C10 ratio – elevated.	
<b>Prenatal testing:</b>	Yes, DNA and enzymatic.	
<b>Miscellaneous Information:</b>	With borderline C8 elevations, it is possible to detect carriers of MCADD.	
<b>Credit:</b>	<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>	
<b>Sites of Reference:</b>	<b>National Library of Medicine Genetics Home Reference</b> <u>Medium-chain acyl-coenzyme A dehydrogenase deficiency</u> <a href="http://ghr.nlm.nih.gov/condition=mediumchainacylcoenzymeadehydrogenasedeficiency">http://ghr.nlm.nih.gov/condition=mediumchainacylcoenzymeadehydrogenasedeficiency</a>	
Support Groups:	<b>United Mitochondrial Disease Foundation</b> 8085 Saltsburg Road, Suite 201 Pittsburg, PA 15239 412-793-8077 412-793-6477 info@umdf.org <a href="http://www.umdf.org">www.umdf.org</a>  <b>FOD Family Support Group</b> 805 Montrose Drive Greensboro, NC 24710 www.fodsupport.org/ Contact: Deb Lee Gould (336) 547-8682 <a href="mailto:FODgroup@aol.com">FODgroup@aol.com</a>	<b>James William Lazzaro Foundation</b> 4493 Liberty Road South Euclid, OH 44121 www.jwlsite.com/ Contact: Jamie Lazzaro (502) 254-2209 <a href="mailto:info@jwlsite.com">info@jwlsite.com</a>