

Disease Name	
ISOBUTYRYL-CoA DEHYDROGENASE DEFICIENCY (IBD)	
<i>(ACYL-CoA DEHYDROGENASE FAMILY, MEMBER 8)</i>	
Classification:	Fatty acid oxidation defect
Genetic Information	
Inheritance:	Presumed autosomal recessive.
Population Incidence:	Rare, less than five cases.
Ethnic Incidence:	No known population at increased risk.
Gene & Location:	ACAD8 - 11q25
Common Mutation:	No known common mutations.
OMIM #	*604773
Disease Information	
Symptom Onset:	12 months of age
Symptoms:	Initial patient presented with dilated cardiomyopathy, low carnitine and anemia. Was small for age at presentation, but normal growth resumed with treatment. A three-year-old identified, as a newborn, through screening has remained asymptomatic.
Physical Findings:	Cardiomyopathy; no dysmorphisms.
Treatment:	Carnitine therapy reversed the cardiomyopathy. Moderate protein restriction to reduce valine intake and avoidance of fasting is prudent.
Natural History without treatment:	Unknown.
Natural History with treatment:	Improvement in symptoms of cardiomyopathy and anemia with improved growth and normal development.
Metabolic Information	
Missing Enzyme & Location:	ISOBUTYRYL-CoA DEHYDROGENASE- impaired valine metabolism
MS/MS profile:	C4 (butyryl/ isobutyryl carnitine)- isolated elevation.
Prenatal testing:	Enzyme analysis on amniocytes or CVS.
Miscellaneous Information:	Need to differentiate from SCAD- on urine organic acid analysis will NOT see elevated Ethylmalonic acid.
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:	Identification of isobutyryl-CoA dehydrogenase and its deficiency in humans www.faseb.org/genetics/ashg00/f277.htm
Support Groups:	Organic Acidemia Association www.oaanews.org/ 13210 35th Avenue Plymouth, MN 55441 Contact: Kathy Stagni