



Genetic Fact Sheets for Professionals

Organic Acid 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>

Disease name	Isobutyryl-CoA dehydrogenase deficiency
Alternate name(s)	Acyl-CoA dehydrogenase family, member 8
Acronym	N/A
Disease classification	Organic Acid Disorder/Fatty Acid Oxidation Defect
Variants	None
Variant name	N/A
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.
Natural history without treatment	Unknown
Natural history with treatment	Improvement in symptoms of cardiomyopathy and anemia with improved growth and normal development.
Treatment	Moderate protein restriction. Carnitine therapy.
Other	N/A
Physical phenotype	Cardiomyopathy. No dysmorphisms reported.
Inheritance	Presumed autosomal recessive
General population incidence	Rare. Less than five cases reported.
Ethnic differences	None known

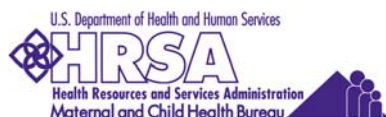
Population	N/A
Ethnic incidence	N/A
Enzyme location	Mitochondria
Enzyme function	Metabolism of valine
Missing enzyme	Isobutyryl-CoA dehydrogenase
Metabolite changes	N/A
Gene	ACAD8
Gene location	11q25
DNA testing available	No
DNA testing detail	No common mutations known.
Prenatal testing	May be possible by enzyme analysis on amniocytes or CVS.
MS/MS profile	C4 butyryl carnitine elevation
OMIM link	www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=60473
Genetests link	None
Support group	Fatty Acid Oxidation Support Network www.fodsupport.org Save Babies through Screening Foundation www.savebabies.org Genetic Alliance www.geneticalliance.org

Document Info

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