

Disease Name:

ISOBUTYRYL-CoA DEHYDROGENASE DEFICIENCY
(ACYL-CoA DEHYDROGENASE FAMILY, MEMBER 8)

Classification:

Fatty acid oxidation defect

Genetic Information:

Inheritance: Presumed autosomal recessive

Population Incidence: Rare, less than 5 cases

Ethnic Incidence: No known population at increased risk

Gene & Location: Acyl-CoA dehydrogenase family, member 8 - 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.

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References:

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