Disease Name: MALONYL-CoA DECARBOXYLASE DEFICIENCY
(MALONIC ACIDURIA)

Classification:
Disorder of ketone metabolism and fatty acid oxidation

Genetic Information:
Inheritance: Autosomal recessive
Population Incidence: Rare, less than 20 reported cases
Ethnic Incidence: No known population at increased risk
Gene & Location: Malonyl-CoA decarboxylase- 16q24
Common Mutation: No known common mutations
OMIM #: #248360

Disease Information:
Symptom Onset: Age of presentation ranges from 3 days to 13 years old.
Symptoms: All patients have had developmental delay and 20-40% have other symptoms, including hypotonia, hypoglycemia, metabolic acidosis, cardiomyopathy (hypertrophic and/or dilated), diarrhea, vomiting, ketosis, seizures, lactic acidemia, microcephaly and low cholesterol.
Physical Findings: Single report of micropenis and renal dysplasia in a patient with malonic aciduria. Another with epicanthal folds and long face.
Treatment: Carnitine, high-carbohydrate diet, decreased long-chain fatty acids in diet and MCT supplement. Efficacy of treatment has not been determined.
Natural History without treatment: One patient died as a neonate and 2 died in infancy. Symptoms tend to be worse with stressors like illness or fasting. A patient in her 20’s has severe cognitive impairment and spastic quadriplegia.
Natural History with treatment: Efficacy unknown.

Metabolic Information:
Missing Enzyme & Location: Enzyme is present in both peroxisomes and mitochondria. Malonyl CoA decarboxylase breaks down malonyl CoA to acetyl CoA
MS/MS profile: C3-DC (malonyl carnitine)- elevated
Prenatal testing: Theoretically possible via enzyme analysis on amniocytes or CVS.
Miscellaneous Information: The malonic acid and malonyl-CoA are thought to be toxic to the brain cells and cause the neurological symptoms.

Prepared for the NW Regional Newborn Screening Program by Sara Copeland MD, Judith Tuerck RN MS and Lorinda Paradise at OHSU in Portland, OR.

References:


16. OMIM- Online Mendelian Inheritance in Man; MALONYL-CoA DECARBOXYLASE DEFICIENCY- #248360.


