

| Disease Name | |
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| MALONYL-CoA DECARBOXYLASE DEFICIENCY (MA) | |
| <i>(MALONIC ACIDURIA)</i> | |
| 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: | MLYCD, MCD genes on 16q24 |
| Common Mutation: | No known common mutations. |
| OMIM # | #248360 |
| Disease Information | |
| Symptom Onset: | Age of presentation ranges from three days to 13 years old. |
| Symptoms: | All patients have had developmental delay and 20-40 percent 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, and decreased fatty acids in diet. Efficacy of treatment has not been determined. |
| Natural History without treatment: | One patient died as a neonate and two 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 quadriparesis. |
| Natural History with treatment: | Unknown, no known prospectively treated patients. |
| 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. |
| 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: | |

Support Groups:

Children Living with Inherited Metabolic Diseases (CLIMB)

Climb Building
176 Nantwich Road
Crewe, CW2 6BG
United Kingdom
(+44) 0870 7700 326
(+44) 0870 7700 327
steve@climb.org.uk
www.climb.org.uk