

Disease Name:

3-METHYLGLUTACONIC ACIDURIA TYPE IV
(MGA, TYPE IV)

Classification:

Organic aciduria

Genetic Information:

Inheritance: Autosomal recessive

Population Incidence: Unknown

Ethnic Incidence: No known population at increased risk

Gene & Location: Unknown, likely many genes involved

Common Mutation: No known common mutations

OMIM # #250951

Disease Information:

Symptom Onset: Presents in the first year of life

Symptoms: Type IV patients have a disparate variety of symptoms with the only commonality being the mild excretion of 3-MGA in the urine. Patients have a clinically heterogeneous and nonspecific presentation and clinical course. There is variable psychomotor retardation; hypertonicity; hypotonia; optic atrophy; dysmorphic features; seizures; cardiomyopathy and hepatic dysfunction. Others have been noted to have neurodegeneration; deafness; failure to thrive; absence of acidosis. Some patients may have Pearson syndrome- hematologic disorder, lactic acidemia, and abnormality of the electron transport chain with identified mitochondrial DNA deletions.

Physical Findings: No dysmorphisms, may have neurological findings

Treatment: There are no effective treatments.

Natural History without treatment: Varies from asymptomatic except for organic aciduria to neurodegeneration and deafness.

Natural History with treatment: Same as for untreated group

Metabolic Information:

Missing Enzyme & Location: The basic enzyme defect is unknown

MS/MS profile: C5-OH methylcrotonyl carnitine - elevated

Prenatal testing: None available, basic biochemical defect is unknown.

**Miscellaneous
Information:**

Prepared for the NW Regional Newborn Screening Program

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