

**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 (3-hydroxyisovaleryl carnitine) - elevated

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

**Miscellaneous  
Information:**

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

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