

Disease Name	
3-METHYLGLUTACONIC ACIDURIA TYPE IV	
<i>(MGA, TYPE IV)</i>	
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.
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:	Some patients may have Pearson syndrome hematological disorder, lactic acidemia, and abnormality of the electron transport chain with identified mitochondrial DNA deletions.
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:	Organic Acidemia Association www.oaanews.org/ 13210 35th Avenue Plymouth, MN 55441 Contact: Kathy Stagni (763) 559-1797 OAANews@aol.com

