

# Health Care Provider Fact Sheet

<b>Disease Name</b>	<b>3-Methylglutaconic Aciduria</b>
<b>Acronym</b>	3 MGA
<b>Disease Classification</b>	Organic Acid Disorder
<b>Description</b>	Name used to describe five different disorders that impair the functioning of the mitochondria. Disruption of mitochondrial function pathways results in increased amounts of 3-methylglutaconic acid and 3-methylglutaric acid.
<b>Symptom onset</b>	Variable, from infancy to early childhood
<b>Symptoms</b>	Symptoms range from minimal to severe. Each type has its own signs and symptoms. Type I = speech delays, psychomotor delays, metabolic acidosis, dystonia, and spastic quadriparesis. Type II= Dilated cardiomyopathy, neutropenia, and skeletal myopathy. Type III = Optic nerve atrophy, cognitive deficit, and choreiform movements. Type IV = variable and overlap with Types I-III. Type V = Dilated cardiomyopathy with ataxia, cryptorchidism, and hypospadias.
<b>Natural History w/o Treatment</b>	Variable. Can be asymptomatic to patients with severe neurological dysfunction, hypoglycemia, and acidosis.
<b>Natural History with Treatment</b>	Same as without treatment. Theoretically, in some types, the expected outcome would be improved neurological status and fewer problems with fasting and illness
<b>Treatment</b>	Treatment is generally supportive and often involves a multidisciplinary team. With 3MGA Type I, if diagnosed presymptomatically, carnitine supplementation and moderate leucine restrictions may be beneficial
<b>Inheritance</b>	Autosomal Recessive
<b>General population incidence</b>	RARE, Fewer than 20 cases of 3-methylglutaconic Aciduria type I have been reported
<b>Missing Enzyme and Location</b>	The basic enzyme defect for types III and IV is unknown. For types I and II the location is thought to be in the mitochondria.
<b>MS/MS Profile</b>	C5-OH elevation
<b>OMIM Link</b>	<a href="http://www.omim.org">www.omim.org</a> ID#s *250950, *258501, *610198, *302060, *250951, *606580
<b>Genetests Link</b>	<a href="http://www.genetes.org">www.genetes.org</a>
<b>Support Group and References</b>	Genetics Home Reference <a href="http://www.ghr.nlm.nih.gov/ghr/page/home">www.ghr.nlm.nih.gov/ghr/page/home</a> Gene Reviews <a href="http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&amp;part=mga3">http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&amp;part=mga3</a>