



Genetic Fact Sheets for Professionals

Organic Acid Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – [http:// www.newbornscreening.info](http://www.newbornscreening.info)

Disease name	3-hydroxy-3-methylglutaryl-CoA lyase deficiency
Alternate name(s)	Hydroxymethylglutaric aciduria
Acronym	HMG-CoA lyase deficiency
Disease classification	Organic Acid Disorder
Variants	No
Variant name	N/A
Symptom onset	Infancy (6 months to 2 years)
Symptoms	Persistent vomiting, lethargy, hypotonia, coma, seizures, apnea, hepatomegaly.
Natural history without treatment	Recurrent episodes of acute illness usually in response to fasting or to viral infection. Any episode can lead to death or developmental delay if severe enough.
Natural history with treatment	Normal IQ and development are possible. Severe hypoglycemic episodes may result in seizures and mental retardation.
Treatment	Avoidance of fasting. Low fat, protein and high carbohydrate diet. Cornstarch supplementation. Carnitine supplementation. Intravenous glucose to treat hypoglycemia during crisis episodes.
Other	Crises consist of severe acidosis and hypoglycemia treated with IV glucose and bicarbonate administration.
Physical phenotype	Possible microcephaly
Inheritance	Autosomal recessive
General population incidence	Rare
Ethnic differences	No

Population	N/A
Ethnic incidence	N/A
Enzyme location	Liver, fibroblasts and leukocytes
Enzyme function	Catalyzes the final step of leucine degradation and plays a role in ketone formation.
Missing enzyme	HMG CoA lyase
Metabolite changes	3-hydroxy-3-methylglutaric acid in urine, increased levels of glutaric and adipic acids may be elevated in urine during crisis, notable absence of ketosis.
Gene	HMGCL
Gene location	1pter-p33
DNA testing available	No
DNA testing detail	N/A
Prenatal testing	Prenatal testing has been accomplished by analysis of metabolites in maternal urine at 23 weeks. Enzyme is active in amniocytes and prenatal testing should be possible using this method.
MS/MS profile	N/A
OMIM link	www.ncbi.nlm.nih.gov/htbin-post/Omim/dispim?246450
Genetests link	www.genetests.org/servlet/access?prg=j&db=genestar&site=&fcn=d&id=12600&qry=22675&res=nous&res=nointl&key=hp5d0Ly2h80eI&show_flag=c
Support group	Organic Acidemia Association www.oaanews.org Save Babies through Screening Foundation www.savebabies.org Genetic Alliance www.geneticalliance.org

Document Info

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