

Glutaric aciduria type I

Disorder information

Name

Glutaric aciduria type I

Alternative names

Glutaric acidemia type I

Glutaryl-CoA dehydrogenase deficiency

Disease abbreviation

GA-I

OMIM

<http://www.omim.org/entry/231670>

GeneReviews

<http://www.ncbi.nlm.nih.gov/books/NBK1155>

Gene information

Gene symbol

GCDH

Protein name

Glutaryl-CoA dehydrogenase

HGNC gene symbol

GCDH

HGNC gene name

glutaryl-CoA dehydrogenase

Chromosomal location

19p13.2

Genetic Testing Registry

[http://www.ncbi.nlm.nih.gov/gtr/tests/?term=GCDH\[gene\]](http://www.ncbi.nlm.nih.gov/gtr/tests/?term=GCDH[gene])

NCBI Gene

<https://www.ncbi.nlm.nih.gov/gene/2639>

Gene Card

<http://www.genecards.org/cgi-bin/carddisp.pl?gene=GCDH>

Uniprot

<http://www.uniprot.org/uniprot/Q92947>

KEGG

http://www.genome.jp/dbget-bin/www_bget?hsa:2639

Clinical Symptoms

Symptoms that are characteristic of this disorder are highlighted and marked in bold.

Symptom	Neonatal (birth-1mth)	Infancy (1-18mths)	Childhood (1.5-11yrs)	Adolescence (11-16yrs)	Adulthood (>16yrs)
Chorea	?	±	±	±	±
Hypokinesia	?	±	±	±	±
Macrocephaly	±	+	±	±	±
Abasia	?	±	±	±	±
Astasia	?	?	±	±	±
Ataxia	?	?	±	±	±
Axial hypotonia	±	±	±	?	?
Dysarthria	?	±	±	±	±
Encephalopathic crisis, acute	?	±	±	?	?
Feeding difficulties	?	±	±	±	±
Headache	?	?	?	±	±
Other extrastriatal abnormalities	?	±	±	+	+
Periventricular white matter abnormalities	?	±	±	+	+
Pneumonia	?	±	±	±	±
Spasticity	?	?	±	±	±
Striatal atrophy	?	±	±	±	±
Swallowing difficulties	?	±	±	±	±
Temporal hypoplasia, dilated external CSF spaces	+	+	±	±	±
Vertigo	?	?	?	±	±
Vomiting	?	±	±	±	±

Biochemical markers

Markers that are characteristic of this disorder are highlighted and marked in bold.

Test	Symptom	HMDB	Neonatal (birth-1mth)	Infancy (1-18mths)	Childhood (1.5-11yrs)	Adolescence (11-16yrs)	Adulthood (>16yrs)
Carnitine and acylcarnitines	C5-DC Glutarylcarnitine (blood)	HMDB001313 0	n-↑	n-↑	n-↑	n-↑	n-↑
	C5-DC Glutarylcarnitine (plasma)	HMDB001313 0	n-↑	n-↑	n-↑	n-↑	n-↑
Organic acids	3-Hydroxyglutaric acid (urine)	HMDB00428	n-↑	n-↑	n-↑	n-↑	n-↑
	Glutaric acid (plasma)	HMDB00661	n-↑	n-↑	n-↑	n-↑	n-↑
	Glutaric acid (urine)	HMDB00661	n-↑	n-↑	n-↑	n-↑	n-↑
	Glutaconic acid (urine)	HMDB00620	n-↑	n-↑	n-↑	n-↑	n-↑
Routine tests	ASAT/ALAT (plasma)	HMDBP08982; HMDBP00284; HMDBP00283; HMDBP00850; HMDBP03670	?	↑	↑	↑	↑
	Creatine kinase (plasma)	HMDBP00720; HMDBP00722; HMDBP00719; HMDBP00721	?	↑	↑	↑	↑
No Test Specified	Glutaric acid (dried blood spot)	HMDB00661	n-↑	n-↑	n-↑	n-↑	n-↑