

Disease

Glutaric aciduria, type I^{1, 2}

Gene

GCDH

Inheritance	AR
Current Treatments	avoid fasting, carnitine, protein restricted diet, restrict lysine, hydroxylysine, and tryptophan
Evidence for Treatment	guideline ^{3, 4}
Non-Molecular Testing Availability	yes
Non-Molecular Test	urine organic acids, leukocyte enzyme activity ⁵
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC142
Disease Frequency (per 100,000)	1.06 ⁶
Genes Included in Disease Frequency	GCDH
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Glutaric+aciduria&term=&cntry=&state=&city=&dist=#

References

- ¹ <https://www.omim.org/entry/231670>
- ² <https://pubmed.ncbi.nlm.nih.gov/33069577/>
- ³ <https://www.ncbi.nlm.nih.gov/pubmed/27853989>
- ⁴ http://www.bimdg.org.uk/store/guidelines/ER-GA1-v4_549337_05042017.pdf
- ⁵ <https://www.ncbi.nlm.nih.gov/books/NBK546575/>
- ⁶ <https://www.ncbi.nlm.nih.gov/pubmed/25022222>