Rx-Genes
Treatment Guidance at the Point of Care

Disease

Glutaric acidemia IIA[1, 2, 3]

Gene

ETFA

Inheritance	AR
Current Treatments	riboflavin, carnitine, glycine, Coenzyme Q10 supplementation, fat restriction, avoidance of fasting, and a diet rich in carbohydrates, D,L-3-hydroxybutyrate
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	Plasma acylcarnitine profile, Urine organic acid analysis, [1]
Disease Frequency (per 100,000)	0.4[4]
Genes Included in Disease Frequency	ETFA, ETFB, ETFDH, FLAD1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Glutaric+Acidemia+II&term=&cntry=&state=&city=&dist=

References

[1] https://www.ncbi.nlm.nih.gov/books/NBK558236/

[2] https://pubmed.ncbi.nlm.nih.gov/31331668/

[3] https://www.ncbi.nlm.nih.gov/pubmed/31904027

[4] https://www.ncbi.nlm.nih.gov/pubmed/22766612