

Disease

Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency^{1, 2}

Gene

FLAD1

Inheritance	AR
Current Treatments	riboflavin, carnitine, glycine, Coenzyme Q10 supplementation, fat restriction, avoidance of fasting, and a diet rich in carbohydrates
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	Plasma acylcarnitine profile, Urine organic acid analysis ³
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=flad1&term=&cntry=&state=&city=&dist=

References

¹ <https://pubmed.ncbi.nlm.nih.gov/31392824/>

² <https://omim.org/entry/255100>

³ <https://pubmed.ncbi.nlm.nih.gov/33279678/>

⁴ <https://www.ncbi.nlm.nih.gov/pubmed/22766612>