

Disease

Very long-chain acyl-CoA dehydrogenase deficiency^{1, 2, 3}

Gene

ACADVL

Inheritance	AR
Current Treatments	avoid fasting, carnitine, restrict LCFA, bezafibrate, triheptanoin
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	no
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC126
Disease Frequency (per 100,000)	1.665 ⁵
Genes Included in Disease Frequency	ACADVL
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=ACADVL&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK6816/>
- ² <https://pubmed.ncbi.nlm.nih.gov/31372341/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/32885845/>
- ⁴ <https://www.sciencedirect.com/science/article/abs/pii/S1096719220302018>
- ⁵ <https://ghr.nlm.nih.gov/condition/very-long-chain-acyl-coa-dehydrogenase-deficiency#statistics>