

Disease

Medium-chain acyl-CoA dehydrogenase deficiency¹

Gene

ACADM

Inheritance	AR
Current Treatments	avoid fasting
Evidence for Treatment	guideline ^{2, 3}
Non-Molecular Testing Availability	yes
Non-Molecular Test	Urine acylglycine analysis ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC1016
Disease Frequency (per 100,000)	9.35 ⁴
Genes Included in Disease Frequency	ACADM
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=acadm&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1424/>
- ² <https://pediatrics.aappublications.org/content/early/2020/12/24/peds.2020-040303>
- ³ http://www.bimdg.org.uk/store/guidelines/MCADD_Dietary_BIMDG_babies_2015v0_7_742847_13052015.pdf
- ⁴ <https://ghr.nlm.nih.gov/condition/medium-chain-acyl-coa-dehydrogenase-deficiency#statistics>