

Disease

Alpha-methylacetoacetic aciduria¹

Gene

ACAT1

Inheritance	AR
Current Treatments	avoid fasting, carnitine, riboflavin, protein restricted diet
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	urine organic acids, plasma acylcarnitine profile ²
Disease Frequency (per 100,000)	0.415 ³
Genes Included in Disease Frequency	ACAT1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=ACAT1&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/30393371>

² <https://pubmed.ncbi.nlm.nih.gov/30393371/>

³ <https://www.ncbi.nlm.nih.gov/pubmed/28726122>