

Disease

3-Hydroxy-3-methylglutaryl coenzyme A lyase deficiency^{1,2}

Gene

HMGCL

Inheritance	AR
Current Treatments	IV glucose during acute episodes, avoid fasting, carnitine, protein restricted diet
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	urine organic acids, plasma acylcarnitine profile ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=HMGCL&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/28583327>

² <https://pubmed.ncbi.nlm.nih.gov/32059735/>