

Disease

3-hydroxy-3-methylglutaryl-CoA synthase deficiency^{1, 2, 3}

Gene

HMGCS2

Inheritance	AR
Current Treatments	IV glucose during acute episodes, avoid prolonged fasting
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=HMGCS2&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/605911>
- ² <https://www.sciencedirect.com/science/article/abs/pii/S0009898120301509>
- ³ <https://pubmed.ncbi.nlm.nih.gov/40515583/>