

Disease

Trifunctional protein deficiency^{1, 2, 3}

Gene

HADHB

Inheritance	AR
Current Treatments	IV glucose during acute episodes, avoid fasting, carnitine, restrict LCFA, bezafibrate, triheptanoin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
Disease Frequency (per 100,000)	1.16 ⁴
Genes Included in Disease Frequency	HADHA and HADHB
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Trifunctional+protein+deficiency&term=&cntry=&state=&city=&

References

- ¹ <https://omim.org/entry/143450>
- ² <https://www.sciencedirect.com/science/article/pii/S2214426920300562>
- ³ <https://pubmed.ncbi.nlm.nih.gov/32885845/>
- ⁴ <https://ghr.nlm.nih.gov/condition/long-chain-3-hydroxyacyl-coa-dehydrogenase-deficiency#statistics>