

Disease

Mitochondrial short-chain enoyl-CoA hydratase-1 deficiency^{1, 2}

Gene

ECHS1

Inheritance	AR
Current Treatments	valine restricted diet
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	acylcarnitine profile ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=ECHS1&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/616277>

² <https://pubmed.ncbi.nlm.nih.gov/32642440/>