

Disease

Holocarboxylase synthetase deficiency^{1, 2}

Gene

HLCS

Inheritance	AR
Current Treatments	biotin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	urine organic acids ³
Disease Frequency (per 100,000)	1.15 ⁴
Genes Included in Disease Frequency	HLCS
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Holocarboxylase+synthetase&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/253270>

² <https://www.babysfirsttest.org/newborn-screening/conditions/holocarboxylase-synthetase-deficiency>

³ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4832086/>

⁴ <https://ghr.nlm.nih.gov/condition/holocarboxylase-synthetase-deficiency#statistics>