

Disease

Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase^{1, 2, 3}

Gene

AHCY

Inheritance	AR
Current Treatments	methionine-restricting diet and supplementation with creatine and phosphatidylcholine. Liver transplantation
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum amino acids, serum CK ⁴
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Hypermethioninemia+with+S-Adenosylhomocysteine+Hydrolase+Deficiency&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/613752>
- ² <https://pubmed.ncbi.nlm.nih.gov/26974671/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/26095522/>
- ⁴ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4790936/>