

Disease

Homocystinuria-megaloblastic anemia, cbl E type^{1, 2}

Gene

MTRR

Inheritance	AR
Current Treatments	hydroxocobalamin, folic acid, betaine,
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum amino acids, urine organic acids ³
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=mtrr&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/236270>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/25526710>
- ³ <https://www.ncbi.nlm.nih.gov/books/NBK1328/#cbl.Diagnosis>