

Disease

Methylenetetrahydrofolate reductase deficiency^{1, 2}

Gene

MTHFR

Inheritance	AR
Current Treatments	betaine, 5-methyltetrahydrofolate
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma homocysteine, methionine levels and CSF 5-methyltetrahydrofolate level ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=MTHFR&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/236250>

² <https://www.ncbi.nlm.nih.gov/pubmed/26898294>