

Disease

Folate dependent megaloblastic anemia¹

Gene

SLC19A1

Inheritance	AR
Current Treatments	folic acid
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma amino acids for homocysteine and sarcosine levels and urine 5-amino-4-imidazolecarboxamide riboside ¹
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SLC19A1&term=&cntry=&state=&city=&dist=

References

¹ <https://pubmed.ncbi.nlm.nih.gov/32276275/>