

Disease

Cerebral folate transport deficiency¹

Gene

FOLR1

Inheritance	AR
Current Treatments	folinic acid
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	cerebrospinal fluid 5-methyltetrahydrofolate level ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=folr1&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/613068>

² <https://pubmed-ncbi-nlm-nih-gov.ezproxy3.lhl.uab.edu/33105619/>