

Disease

Hereditary folate malabsorption¹

Gene

SLC46A1

Inheritance	AR
Current Treatments	5-formyltetrahydrofolate (5-formylTHF, folinic acid, Leucovorin) or the active isomer of 5-formylTHF (Isovorin or Fusilev) Parenteral (intramuscular) or high-dose oral
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	CSF and serum folate levels ²
Disease Frequency (per 100,000)	1.72 ³
Genes Included in Disease Frequency	JAK3, IL2RG, LCK, NHEJ1, FOXN1, PNP, PRKDC, ORAI1, IL7R, PTPRC, ADA, RAG1, RAG2, DCLRE1C, CD3D, CD3E, CD247, AK2, CORO1A, CHD7, CIITA, RFXANK, RFX5, RFXAP, DOCK8, DOCK2, LIG4, MTHFD1, NBN, SLC46A1, PGM3, RMRP, STIM1, TTC7A, ZAP70, RAC2, PAX1, LAT, NUDCD3
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SLC46A1&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1673/#folate-mal.Summary>

² <https://www.ncbi.nlm.nih.gov/books/NBK1673/>

³ www.mdpi.com/2409-515X/3/2/15/pdf