

Disease

Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia^{1, 2}

Gene

MTHFD1

Inheritance	AR
Current Treatments	hydroxocobalamin, folinic acid and betaine
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	T and B Lymphocyte and Natural Killer Cell Profile, complete blood count with MCV, plasma homocysteine and methylmalonic acid levels, CSF 5-methyltetrahydrofolate level ³
Disease Frequency (per 100,000)	1.72 ⁴
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=MTHFD1&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/27707659>
- ² <https://pubmed.ncbi.nlm.nih.gov/32414565/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/30916789/>
- ⁴ www.mdpi.com/2409-515X/3/2/15/pdf