

Disease

Transcobalamin receptor deficiency^{1, 2}

Gene

CD320

Inheritance	AR
Current Treatments	cobalamin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum amino acids, urine organic acids ¹
Disease Frequency (per 100,000)	1.5 ³
Genes Included in Disease Frequency	MMUT, MMAA, MMAB, MMADHC, MMACHC, MCEE, PRDX1, HCFC1, THAP11, ZNF143, LMBRD1, ABCD4, CD320
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=cd320&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/613646>

² <https://pubmed.ncbi.nlm.nih.gov/29663633/>

³ <https://ghr.nlm.nih.gov/condition/methylmalonic-acidemia#statistics>