

Disease

Transcobalamin II deficiency^{1, 2, 3}

Gene

TCN2

Inheritance	AR
Current Treatments	cobalamin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum amino acids, vitamin B12, and methylmalonic acid levels ⁴
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Transcobalamin&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/275350>
- ² <https://pubmed.ncbi.nlm.nih.gov/32841161/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/33685478/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/25914105/>