

Disease

# Transcobalamin receptor deficiency<sup>1, 2</sup>

Gene

## CD320

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

References

<sup>1</sup> <https://omim.org/entry/613646>

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

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