

## Disease

# Methylmalonic acidemia, cblB type<sup>1, 2</sup>

## Gene

### MMAB

|                                                 |                                                                                                                                                                                                                                                             |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                                                          |
| <b>Current Treatments</b>                       | cobalamin, N-carbamylglutamate, carnitine & diet                                                                                                                                                                                                            |
| <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>3</sup>                                                                                                                                                                                                         |
| <b>ClinGen</b>                                  | <a href="https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC122">https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC122</a>                                                                           |
| <b>Disease Frequency (per 100,000)</b>          | 1.5 <sup>4</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=Methylmalonic+Acidemia&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=Methylmalonic+Acidemia&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

## References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1328/>
- <sup>2</sup> <https://pubmed-ncbi-nlm-nih-gov.ezproxy3.lhl.uab.edu/33552909/>
- <sup>3</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1328/#cbl.Diagnosis>
- <sup>4</sup> <https://ghr.nlm.nih.gov/condition/methylmalonic-acidemia#statistics>