

Disease

Methylmalonic acidemia, mut(0) type^{1, 2, 3}

Gene

MMUT

Inheritance	AR
Current Treatments	cobalamin, N-carbamylglutamate, carnitine, diet, liver transplant
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum amino acids, urine organic acids ⁵
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC122
Disease Frequency (per 100,000)	1.5 ⁶
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=Methylmalonic+Acidemia&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/251000>
- ² <https://www.ncbi.nlm.nih.gov/books/NBK1231/#top>
- ³ <https://pubmed.ncbi.nlm.nih.gov/36331064/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/33595124/>
- ⁵ <https://www.ncbi.nlm.nih.gov/books/NBK1231/#mma.Diagnosis>
- ⁶ <https://ghr.nlm.nih.gov/condition/methylmalonic-acidemia#statistics>