Rx-Genes
Treatment Guidance at the Point of Care

Disease

Combined methylmalonic acidemia and homocystinuria, cblD type[1]

Gene

MMADHC

Inheritance	AR
Current Treatments	cobalamin, carnitine & diet
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum amino acids, urine organic acids[2]
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC122
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=Methylmalonic+Acidemia&term=&cntry=&state=&city=&dist=

References

[1] https://www.ncbi.nlm.nih.gov/books/NBK1328/

[2] https://www.ncbi.nlm.nih.gov/books/NBK1328/#cbl.Diagnosis

[3] https://ghr.nlm.nih.gov/condition/methylmalonic-acidemia#statistics