Rx-Genes
Treatment Guidance at the Point of Care

**Disease**

# Combined methylmalonic acidemia and homocystinuria, cblX type[1, 2]

**Gene**

## HCFC1

| | |
|---|---|
| **Inheritance** | XL |
| **Current Treatments** | cobalamin, carnitine & diet |
| **Evidence for Treatment** | expert opinion |
| **Non-Molecular Testing Availability** | yes |
| **Non-Molecular Test** | serum amino acids, urine organic acids[3] |
| **Disease Frequency (per 100,000)** | 1.5[4] |
| **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://pubmed.ncbi.nlm.nih.gov/36207831/

3 https://www.ncbi.nlm.nih.gov/books/NBK1328/#cbl.Diagnosis

4 https://ghr.nlm.nih.gov/condition/methylmalonic-acidemia#statistics