

## 32-4218: Recombinant Human Methylmalonic Aciduria cblD type, with Homocystinuria

**Alternative Name :** Chromosome 2 Open Reading Frame 25, Methylmalonic Aciduria (Cobalamin Deficiency) CblD Type With Homocystinuria, Methylmalonic Aciduria And Homocystinuria Type D Protein Mitochondrial, Protein C2orf25 Mitochondrial, CL25022, C2orf25, cblD.

### Description

Source : Escherichia Coli. MMADHC Human Recombinant produced in E.coli is a single, non-glycosylated polypeptide chain containing 281 amino acids (39-296) and having a molecular mass of 31.0 kDa. MMADHC is fused to a 23 amino acid His-tag at N-terminus. MMADHC is a mitochondrial protein which takes part in an early step of vitamin B12 metabolism. Vitamin B12 (cobalamin) is vital for regular development and existence in humans. Mutations in MMADHC can result in methylmalonic aciduria and homocystinuria type cblD, a cobalamin metabolism syndrome which is characterized by decreased levels of the coenzymes methylcobalamin and adenosylcobalamin.

### Product Info

**Amount :** 20 µg  
**Purification :** Greater than 90% as determined by SDS-PAGE.  
**Content :** The MMADHC solution (1mg/ml) contains 20mM Tris-HCl buffer (pH 8.0), 0.15M NaCl, 1mM DTT and 10% glycerol.  
**Storage condition :** Store at 4°C if entire vial will be used within 2-4 weeks. Store, frozen at -20°C for longer periods of time. For long term storage it is recommended to add a carrier protein (0.1% HSA or BSA). Avoid multiple freeze-thaw cycles.  
**Amino Acid :** MGSSHHHHHH SSGLVPRGSH MGSSDESHVA AAPPDICSRT VWPDETMGPF GPQDQRFQLP  
 GNIGFDCHLN GTASQKKSLV HKTLPDVLAELPSSERHEFV MAQYVNEFQG NDAPVEQEIN SAETYFESAR  
 VECAIQTCP ELLRKDFESLF PEVANGKLMI LTVTQTKND MTWVSEEEVEI EREVLEKFI NGAKEICYAL  
 RAEGYWADFI DPSSGLAFFG PYTNNTLFET DERYRHLGFS VDDLGCCKVI RHSLWGTHVV VGSIFTNATP  
 DSHIMKKLSG N

