

Recombinant Human methylmalonic aciduria (cobalamin deficiency) cbID type, with homocystinuria, His-tagged

Cat. No. MMADHC-432H Lot. No. (See product label)

SPECIFICATION

Product Overview	MMADHC, 39-296aa, Human, His tag, E.coli
Species	Human
Source	E.coli
ProteinLength	39-296 a.a.
Description	MMADHC is a mitochondrial protein that is involved in an early step of vitamin B12 metabolism. Vitamin B12 (cobalamin) is essential for normal development and survival in humans. Mutations in this gene cause methylmalonic aciduria and homocystinuria type cbID (MMADHC), a disorder of cobalamin metabolism that is characterized by decreased levels of the coenzymes adenosylcobalamin and methylcobalamin. Recombinant human MMADHC protein, fused to His-tag at N-terminus, was expressed in E.coli and purified by using conventional chromatography techniques.
Form	Liquid. In 20mM Tris-HCl buffer (pH 8.0) containing 0.15M NaCl, 10% glycerol, 1mM DTT.
Molecular Mass	31 kDa (281aa) confirmed by MALDI-TOF
AA Sequence	MGSSHHHHHH SSSLVPRGSH MGSSDESHVA AAPPDICSRT VWPDETMGPF GPQDQRFQLP GNIGFDCHLN GTASQKKSLV HKTLPDVLAE PLSSERHEFV

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MAQYVNEFQG NDAPVEQEIN SAETYFESAR VECAIQTCP E LLRKDFESLF
 PEVANGKLMI LTVTQKTKND MTVWSEEVEI EREVLLEKFI NGAKEICYAL
 RAEGYWADFI DPSSGLAFFG PYTNNTLFET DERYRHLGFS VDDLGCCKVI
 RHSLWGTHVV VGSIFTNATP DSHIMKKLSG N

Purity >90% by SDS - PAGE

Storage Can be stored at +4C short term (1-2 weeks). For long term storage, aliquot and store at -20C or -70C. Avoid repeated freezing and thawing cycles.

Concentration 1 mg/ml (determined by Bradford assay)

GENE INFORMATION

Gene Name MMADHC methylmalonic aciduria (cobalamin deficiency) cblD type, with homocystinuria [*Homo sapiens*]

Official Symbol MMADHC

Synonyms MMADHC; methylmalonic aciduria (cobalamin deficiency) cblD type, with homocystinuria; C2orf25, chromosome 2 open reading frame 25; methylmalonic aciduria and homocystinuria type D protein, mitochondrial; cblD; CL25022; protein C2orf25, mitochondrial; C2orf25;

Gene ID [27249](#)

mRNA Refseq [NM_015702](#)

Protein Refseq [NP_056517](#)

MIM [611935](#)

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UniProt ID Q9H3L0

**Chromosome
Location** 2q23

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