

Recombinant Human Ethylmalonic Aciduria (cobalamin deficiency) cbIC Type, with Homocystinuria, His-tagged

Cat. No. MMACHC-6797H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant humanMMACHC protein, fused to His-tag at N-terminus, was expressed in E.coli andpurified by using conventional chromatography techniques
Species	Human
Source	E.coli
Description	The exact functionof MMACHC is not known, however, its C-terminal region shows similarity toTonB, a bacterial protein involved in energy transduction for cobalamin(vitamin B12) uptake. Hence, it is postulated that this protein may have arole in the binding and intracellular trafficking of cobalamin. Mutations inthis protein are associated with methylmalonic aciduria and homocystinuriatype cbIC
Form	Liquid. In 20mMTris-HCl buffer (pH 8.0) containing 0.15M NaCl, 10% glycerol, 1mM DTT
Molecular Weight	34 kDa (306aa),confirmed by MALDI-TOF
Purity	> 90% by SDS -PAGE
Concentration	0.5 mg/ml(determined by Bradford assay)
Sequences of aminoacids	MGSSHHHHHHSSGLVPRGSH MGSHEPKVA ELKQKIEDTL CPFGFEVYPF QVAWYNELL PAFHLPLPGP TLAFLVLSTPAMFDRLKPF LQSCHLRMLT

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DPVDQCVAYH LGRVRESLPE LQIEIIADYE VHPNRRPKIL
 AQTAHVAGAAAYYQRQDVE ADPWGNQRIS GVCIHPRFGG WFAIRGVLL
 PGIEVPDLPP RKPHDCVPTR ADRIALLEGFNFHWRDWTYR DAVTPQERY
 EEQKAYFSTP PAQRLALLGL AQPSEKPSSP SPDLPFTTPA
 PKKPGNPSRARSWLSPRVSP PASPGP

Storage

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

GENE INFORMATION
Gene Name

MMACHC methylmalonic aciduria (cobalamin deficiency) cblC type, with homocystinuria [Homo sapiens]

Official Symbol

MMACHC

Synonyms

MMACHC; methylmalonic aciduria (cobalamin deficiency) cblC type, with homocystinuria; methylmalonic aciduria and homocystinuria type C protein; cblC; DKFZP564I122; cblC; 1810037K07Rik; BOS_3654; DKFZp564I122; FLJ25671; Methylmalonic aciduria (cobalamin deficiency) cblC type with homocystinuria; Methylmalonic aciduria and homocystinuria type C protein; Methylmalonic aciduria and homocystinuria type C protein homolog; MGC134307; OTTHUMP00000009243; RP11-291L19.3; RP23-177C18.3; RP11-291L19.3

Gene ID

25974

mRNA Refseq

NM_015506

Protein Refseq

NP_056321

MIM

609831

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Uniprot ID	Q9Y4U1
Chromosome Location	1p34.1
Pathway	Vitamin digestion and absorption,organism-specific biosystem; Vitamin digestion and absorption, conservedbiosystem
Function	cobalamin binding

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