

## 32-4217: Recombinant Human Methylmalonic Aciduria cbIC type, with Homocystinuria

**Alternative Name :** Methylmalonic aciduria and homocystinuria type C protein, MMACHC, cbIC, RP11-291L19.3.

### Description

Source : Escherichia Coli. MMACHC Human Recombinant produced in E.Coli is a single, non-glycosylated polypeptide chain containing 306 amino acids (1-282 a.a) and having a molecular mass of 34kDa. MMACHC is fused to a 24 amino acid His-tag at N-terminus & purified by proprietary chromatographic techniques. Though the exact role of MMACHC is unknown, its C-terminal region shows similarity to TonB, which is a bacterial protein involved in energy transduction for cobalamin (vitamin B12) uptake. Therefore, it is suggested that MMACHC may have a role in the binding and intracellular trafficking of cobalamin. MMACHC mutations are linked with methylmalonic aciduria and homocystinuria type cbIC. MMACHC is widely expressed, it is expressed at higher level in the fetal liver, however it is also expressed in the spleen, lymph node, thymus and bone marrow. MMACHC is weakly or not expressed in the peripheral blood leukocytes.

### Product Info

<b>Amount :</b>	20 µg
<b>Purification :</b>	Greater than 90.0% as determined by SDS-PAGE.
<b>Content :</b>	MMACHC protein solution (0.5mg/ml) containing 20mM Tris-HCl buffer (pH 8.0), 0.15M NaCl, 10% glycerol and 1mM DTT.
<b>Storage condition :</b>	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.
<b>Amino Acid :</b>	MGSSHHHHHH SGLVPRGSH MGSHEPKVA ELKQKIEDTL CPGFEVYYPF QVAWYNELLP PAFHLPLPGP TLAFLVLSTP AMFDRLKPF LQSCHLRMLT DPVDQCVAYH LGRVRESLPE LQIEIADYE VHPNRRPKIL AQTAHVAGA AYYYQRQDVE ADPWGNQRIS GVCIHPRFGG WFAIRGVVLL PGIEVPDLPP RKP HDCVPTR ADRIALLEGF NFHWRDWTYR DAVTPQERYE EEQKAYFSTP PAQRLALLGL AQPSEKPSPP SPDLPFTTPA PKKPGNPSRA RSWLSRVSP PASPGP.

