9051 Mira Mesa Blvd, #910094
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# MMACHC Human

*Methylmalonic Aciduria cblC type, with Homocystinuria Human Recombinant*
*RCP1375*

## Product Overview

| | |
|---|---|
| Name | MMACHC Human |
| **Description** | |
| Methylmalonic Aciduria cblC type, with Homocystinuria Human Recombinant | |
| Accession (Primary) | Q9Y4U1 |
| **Synonyms** | |
| Methylmalonic aciduria and homocystinuria type C protein, MMACHC, cblC, RP11-291L19.3. | |
| **Introduction** | |
| 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 cblC. 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. | |
| **Source** | |
| Escherichia Coli. | |
| **Physical Appearance** | |
| Sterile Filtered colorless solution. | |
| **Formulation** | |
| MMACHC protein solution (0.5mg/ml) containing 20mM Tris-HCl buffer (pH 8.0), 0.15M NaCl, 10% glycerol and 1mM DTT. | |
| **Stability** | |
| 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. | |
| **Purity** | |
| Greater than 90.0% as determined by SDS-PAGE. | |
| **Amino acid sequence** | |
| MGSSHHHHHH SSGLVPRGSH MGSH MEPKVA ELKQKIEDTL CPFGFEVYPF QVAWYNELLP PAFHLPLPGP TLAFLVLSTP AMFDRALKPF LQSCHLRMLT DPVDQCVAYH LGRVRESLPE LQIEIIADYE VHPNRRPKIL | |

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AQTAAHVAGA AYYYQRQDVE ADPWGNQRIS GVCIHPRFGG WFAIRGVVLL PGIEVPDLPP RKPHDCVPTR ADRIALLEGF NFHWRDWTYR DAVTPQERYS EEQKAYFSTP PAQRLALLGL AQPSEKPSSP SPDLPFTTPA PKKPGNPSRA RSWLSPRVSP PASPGP.

**Precautions**

MMACHC Human is for research use only and not for use in diagnostic or therapeutic procedures.

## Target Information: ( Q9Y4U1 )