

Recombinant Human MMADHC Protein, Myc/DDK-tagged, C13 and N15-labeled

Cat. No. MMADHC-3429H **Lot. No.** (See product label)

SPECIFICATION

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| Product Overview | MMADHC MS Standard C13 and N15-labeled recombinant protein (NP_056517) with a C-terminal MYC/DDK tag, was expressed in HEK293 cells. |
| Species | Human |
| Source | HEK293 |
| Description | This gene encodes 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. Pseudogenes have been identified on chromosomes 11 and X. |
| Molecular Mass | 32.9 kDa |
| AA Sequence | MANVLCNRARLVSYLPGFCSLVKRVVNPKAFSTAGSSGSDESHVAAAPPDICSRTV WPDETMGPFQDQRFQLPGNIGFDCHLNGTASQKKS LVHKTLPDVLAEPLSSER HEFVMAQYVNEFQGNDA PVEQEINSAETYFESARVECAIQTCELLRKDFESLFPEV ANGKLMILVTQKTKNDMTVWSEEVEIEREV LLEKFINGAKEICYALRAEGYWADFID PSSGLAFFGPYTNNTLFETDERYRHLGFSVDDLGCCKVIRHSLWGTHVVVGSIFTNA TPDSHIMKKLSGNTRTRPLEQKLISEEDLAANDILDYKDDDDKV |
| Purity | > 80% as determined by SDS-PAGE and Coomassie blue staining |

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|-----------------------|--|
| Stability | Stable for 3 months from receipt of products under proper storage and handling conditions. |
| Storage | Store at -80 centigrade. Avoid repeated freeze-thaw cycles. |
| Concentration | 50 µg/mL as determined by BCA |
| Storage Buffer | 100 mM glycine, 25 mM Tris-HCl, pH 7.3. |

GENE INFORMATION

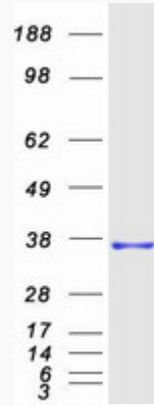
| | |
|------------------------|---|
| Gene Name | MMADHC metabolism of cobalamin associated D [Homo sapiens (human)] |
| Official Symbol | MMADHC |
| Synonyms | MMADHC; methylmalonic aciduria (cobalamin deficiency) cblD type, with homocystinuria; C2orf25, chromosome 2 open reading frame 25; methylmalonic 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 |
| UniProt ID | Q9H3L0 |

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SDS-PAGE



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