

MMACHC Blocking Peptide (C-term)

Synthetic peptide
Catalog # BP20107b

Specification

MMACHC Blocking Peptide (C-term) - Product Information

Primary Accession [Q9Y4U1](#)
Other Accession [NP_056321.2](#)

MMACHC Blocking Peptide (C-term) - Additional Information

Gene ID 25974

Other Names

Methylmalonic aciduria and homocystinuria type C protein, MMACHC

Target/Specificity

The synthetic peptide sequence is selected from aa 254-267 of HUMAN MMACHC

Format

Peptides are lyophilized in a solid powder format. Peptides can be reconstituted in solution using the appropriate buffer as needed.

Storage

Maintain refrigerated at 2-8°C for up to 6 months. For long term storage store at -20°C.

Precautions

This product is for research use only. Not for use in diagnostic or therapeutic procedures.

MMACHC Blocking Peptide (C-term) - Protein Information

Name MMACHC ([HGNC:24525](#))

Function

Cobalamin (vitamin B12) cytosolic chaperone that catalyzes the reductive decyanation of cyanocob(III)alamin (cyanocobalamin, CNCbl) to yield cob(II)alamin and cyanide, using FAD or

MMACHC Blocking Peptide (C-term) - Background

The exact function of the protein encoded by this gene is not known, however, its C-terminal region shows similarity to TonB, a bacterial protein involved in energy transduction for cobalamin (vitamin B12) uptake. Hence, it is postulated that this protein may have a role in the binding and intracellular trafficking of cobalamin. Mutations in this gene are associated with methylmalonic aciduria and homocystinuria type cblC.

MMACHC Blocking Peptide (C-term) - References

Froese, D.S., et al. Mol. Genet. Metab. 100(1):29-36(2010)
Davila, S., et al. Genes Immun. 11(3):232-238(2010)
Profitlich, L.E., et al. Mol. Genet. Metab. 98(4):344-348(2009)
Kim, J., et al. J. Biol. Chem. 284(48):33418-33424(2009)
Richard, E., et al. Hum. Mutat. 30(11):1558-1566(2009)

FMN as cofactors and NADPH as cosubstrate (PubMed:18779575, PubMed:19700356, PubMed:21697092, PubMed:25809485). Cyanocobalamin constitutes the inactive form of vitamin B12 introduced from the diet, and is converted into the active cofactors methylcobalamin (MeCbl) involved in methionine biosynthesis, and 5'-deoxyadenosylcobalamin (AdoCbl) involved in the TCA cycle (PubMed:19801555). Forms a complex with the lysosomal transporter ABCD4 and its chaperone LMBRD1, to transport cobalamin across the lysosomal membrane into the cytosol (PubMed:25535791). The processing of cobalamin in the cytosol occurs in a multiprotein complex composed of at least MMACHC, MMADHC, MTRR (methionine synthase reductase) and MTR (methionine synthase) which may contribute to shuttle safely and efficiently cobalamin towards MTR in order to produce methionine (PubMed:21071249, PubMed:27771510). Also acts as a glutathione transferase by catalyzing the dealkylation of the alkylcob(III)alamins MeCbl and AdoCbl, using the thiolate of glutathione for nucleophilic displacement to generate cob(I)alamin and the corresponding glutathione thioether (PubMed:19801555, PubMed:21697092, PubMed:22642810, PubMed:<a href="http://www.uniprot.org/ci

tations/25809485" target="_blank">25809485). The conversion of incoming MeCbl or AdoCbl into a common intermediate cob(I)alamin is necessary to meet the cellular needs for both cofactors (PubMed:19801555). Cysteine and homocysteine cannot substitute for glutathione in this reaction (PubMed:19801555).

Cellular Location

Cytoplasm, cytosol.

Tissue Location

Widely expressed. Expressed at higher level in fetal liver. Also expressed in spleen, lymph node, thymus and bone marrow. Weakly or not expressed in peripheral blood leukocytes

MMACHC Blocking Peptide (C-term) - Protocols

Provided below are standard protocols that you may find useful for product applications.

- [Blocking Peptides](#)