10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

MMACHC Antibody (C-term)

Affinity Purified Rabbit Polyclonal Antibody (Pab)
Catalog # AP20107b

Product Information

Application	WB, E
Primary Accession	Q9Y4U1
Other Accession	NP_056321.2
Reactivity	Human
Host	Rabbit
Clonality	Polyclonal
Isotype	Rabbit IgG
Clone Names	RB42667
Calculated MW	31728
Antigen Region	239-267

Additional Information

Gene ID	25974
Other Names	Methylmalonic aciduria and homocystinuria type C protein, MMACHC
Target/Specificity	This MMACHC antibody is generated from rabbits immunized with a KLH conjugated synthetic peptide between 239-267 amino acids from the C-terminal region of human MMACHC.
Dilution	WB~~1:1000 E~~Use at an assay dependent concentration.
Format	Purified polyclonal antibody supplied in PBS with 0.09% (W/V) sodium azide. This antibody is purified through a protein A column, followed by peptide affinity purification.
Storage	Maintain refrigerated at 2-8°C for up to 2 weeks. For long term storage store at -20°C in small aliquots to prevent freeze-thaw cycles.
Precautions	MMACHC Antibody (C-term) is for research use only and not for use in diagnostic or therapeutic procedures.

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 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: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

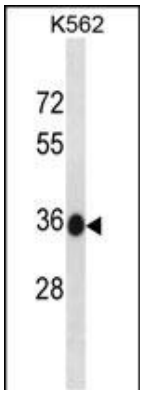
Background

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.

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)

Images



MMACHC Antibody (C-term) (Cat. #AP20107b) western blot analysis in K562 cell line lysates (35ug/lane).This demonstrates the MMACHC antibody detected the MMACHC protein (arrow).

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.