

MUTED Rabbit pAb

Catalog # AP99724

Product Information

Application	IHC-P, IHC-F, IF, E
Primary Accession	Q8TDH9
Other Accession	Q8TDH9
Predicted	Human, Dog, Horse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	21609
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human MUTED
Epitope Specificity	101-187/187
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the Muted family.
SUBUNIT	Interacts with BLOC1S4, DTNBP1/BLOC1S7 and PI4K2A (By similarity). Component of the biogenesis of lysosome-related organelles complex 1 (BLOC-1) composed of BLOC1S1, BLOC1S2, BLOC1S3, BLOC1S4, BLOC1S5, BLOC1S6, DTNBP1/BLOC1S7 and SNAPIN/BLOC1S8. Octamer composed of one copy each BLOC1S1, BLOC1S2, BLOC1S3, BLOC1S4, BLOC1S5, BLOC1S6, DTNBP1/BLOC1S7 and SNAPIN/BLOC1S8. The BLOC-1 complex associates with the AP-3 protein complex and membrane protein cargos. Interacts with BLOC1S6.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a mitochondrial protein that is a member of the solute carrier family. Although this protein was initially thought to be the mitochondrial deoxynucleotide carrier involved in the uptake of deoxynucleotides into the matrix of the mitochondria, further studies have demonstrated that this protein instead functions as the mitochondrial thiamine pyrophosphate carrier, which transports thiamine pyrophosphates into mitochondria. Mutations in this gene cause microcephaly, Amish type, a metabolic disease that results in severe congenital microcephaly, severe 2-ketoglutaric aciduria, and death within the first year. Multiple alternatively spliced variants, encoding the same protein, have been identified for this gene. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	63915
Other Names	BLOS5; HPS11; MU; MUTED; 1810074A19Rik; BL1S5_HUMAN; BLOC1S5; BLOC-1 subunit 5; Protein Muted homolog; BL1S5_MOUSE; biogenesis of lysosomal organelles complex 1 subunit 5; muted homolog (mouse)

Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Name	BLOC1S5 (HGNC:18561)
Synonyms	MUTED
Function	Component of the BLOC-1 complex, a complex that is required for normal biogenesis of lysosome-related organelles (LRO), such as platelet dense granules and melanosomes (PubMed: 32565547). In concert with the AP-3 complex, the BLOC-1 complex is required to target membrane protein cargos into vesicles assembled at cell bodies for delivery into neurites and nerve terminals. The BLOC-1 complex, in association with SNARE proteins, is also proposed to be involved in neurite extension. Plays a role in intracellular vesicle trafficking.

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