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HPS2 Rabbit pAb

Catalog # AP97106

Product Information

Application	WB
Primary Accession	O00203
Other Accession	O00203
Reactivity	Human, Mouse
Predicted	Rat, Pig, Cat, Rabbit, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	121320
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human HPS2
Epitope Specificity	1-100/1094
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 adaptor complexes large subunit family.
Post-translational modifications	Phosphorylated on serine residues.
DISEASE	Defects in AP3B1 are the cause of Hermansky-Pudlak syndrome type 2 (HPS2) [MIM:608233]. Hermansky-Pudlak syndrome (HPS) is a genetically heterogeneous, rare, autosomal recessive disorder characterized by oculocutaneous albinism, bleeding due to platelet storage pool deficiency, and lysosomal storage defects. This syndrome results from defects of diverse cytoplasmic organelles including melanosomes, platelet dense granules and lysosomes. Ceroid storage in the lungs is associated with pulmonary fibrosis, a common cause of premature death in individuals with HPS. HPS2 differs from the other forms of HPS in that it includes immunodeficiency in its phenotype and patients with HPS2 have an increased susceptibility to infections.
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 protein that may play a role in organelle biogenesis associated with melanosomes, platelet dense granules, and lysosomes. The encoded protein is part of the heterotetrameric AP-3 protein complex which interacts with the scaffolding protein clathrin. Mutations in this gene are associated with Hermansky-Pudlak syndrome type 2. Two transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Nov 2012]

Additional Information

Gene ID	8546

Other Names	ADTB3; ADTB3A; HPS; HPS2; PE; AP-3; beta3A; pearl; rim2; XELAEV_18008187mg; ap3b1; AP3B1_BOVIN; Adaptor protein complex AP-3 subunit beta-1; Adaptor-related protein complex 3 subunit beta-1; Beta-3A-adaptin; Clathrin assembly protein complex 3 beta-1 large chain; AP3B1_CANLF; AP3B1_HUMAN; AP3B1_MOUSE; adaptor related protein complex 3 subunit beta 1; adaptor related protein complex 3 beta 1 subunit; beta-3A
Dilution	WB=1:500-2000
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	AP3B1
Synonyms	ADTB3A
Function	Subunit of non-clathrin- and clathrin-associated adaptor protein complex 3 (AP-3) that plays a role in protein sorting in the late-Golgi/trans-Golgi network (TGN) and/or endosomes. The AP complexes mediate both the recruitment of clathrin to membranes and the recognition of sorting signals within the cytosolic tails of transmembrane cargo molecules. AP-3 appears to be involved in the sorting of a subset of transmembrane proteins targeted to lysosomes and lysosome-related organelles. In concert with the BLOC-1 complex, AP-3 is required to target cargos into vesicles assembled at cell bodies for delivery into neurites and nerve terminals.
Cellular Location	Cytoplasmic vesicle, clathrin-coated vesicle membrane; Peripheral membrane protein; Cytoplasmic side. Golgi apparatus. Note=Component of the coat surrounding the cytoplasmic face of coated vesicles located at the Golgi complex
Tissue Location	Ubiquitously expressed.

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