

HPS5 Rabbit pAb

Catalog # AP98025

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q9UPZ3
Other Accession	Q9UPZ3
Reactivity	Mouse
Predicted	Rat, Human, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	127449
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human HPS5
Epitope Specificity	501-600/1129
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 HPS5 family.
SUBUNIT	Component of the biogenesis of lysosome-related organelles complex-2 (or BLOC2) composed of HPS3, HPS5 and HPS6. Directly interacts with HPS6. May interact with all alpha-integrin chains that have an aromatic residue before the first lysine of the conserved KXGFFKR motif, including ITGA2, ITGA3, ITGA5 and ITGA6.
DISEASE	Hermansky-Pudlak syndrome 5 (HPS5) [MIM:614074]: A form of Hermansky-Pudlak syndrome, a genetically heterogeneous 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. Note=The disease is caused by mutations affecting the gene represented in this entry.
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. This protein interacts with Hermansky-Pudlak syndrome 6 protein and may interact with the cytoplasmic domain of integrin, alpha-3. Mutations in this gene are associated with Hermansky-Pudlak syndrome type 5. Multiple transcript variants encoding two distinct isoforms have been identified for this gene. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID 11234

Other Names	AIBP63; BLOC2S2; HPS5_HUMAN; HPS5; Alpha-integrin-binding protein 63; Hermansky-Pudlak syndrome 5 protein; Ruby-eye protein 2 homolog (Ru2); KIAA1017; HPS5 biogenesis of lysosomal organelles complex 2 subunit 2; Hermansky-Pudlak syndrome 5; Ruby-eye protein 2 homolog
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
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	HPS5
Synonyms	AIBP63, KIAA1017
Function	May regulate the synthesis and function of lysosomes and of highly specialized organelles, such as melanosomes and platelet dense granules. Regulates intracellular vesicular trafficking in fibroblasts. May be involved in the regulation of general functions of integrins.
Cellular Location	Cytoplasm, cytosol.
Tissue Location	Widely expressed. Isoform 1:Highly expressed in lungs and testis. Isoform 2:Highly expressed in placenta, kidney, testis ovary, lung and thymus.

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