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HPS1 Polyclonal Antibody

Purified Rabbit Polyclonal Antibody (Pab)
Catalog # AP56679

Product Information

Application	IHC-P, IHC-F, IF, ICC, E
Primary Accession	Q92902
Reactivity	Rat, Bovine
Host	Rabbit
Clonality	Polyclonal
Calculated MW	79292
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human HPS1
Epitope Specificity	501-600/700
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
DISEASE	Defects in HPS1 are the cause of Hermansky-Pudlak syndrome type 1 (HPS1) [MIM:203300]. 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.
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 a component of three different protein complexes termed biogenesis of lysosome-related organelles complex (BLOC)-3, BLOC4, and BLOC5. Mutations in this gene are associated with Hermansky-Pudlak syndrome type 1. Multiple transcript variants encoding distinct isoforms have been identified for this gene; the full-length sequences of some of these have not been determined yet. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	3257
Other Names	Hermansky-Pudlak syndrome 1 protein, HPS1, HPS
Target/Specificity	Ubiquitous.
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC=1:100-500,IF=1:100-500,ELISA=1:5000-10000

Format	0.01M TBS(pH7.4) with 1% BSA, 0.09% (W/V) sodium azide and 50% Glyce
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	HPS1
Synonyms	HPS
Function	Component of the BLOC-3 complex, a complex that acts as a guanine exchange factor (GEF) for RAB32 and RAB38, promotes the exchange of GDP to GTP, converting them from an inactive GDP-bound form into an active GTP-bound form. The BLOC-3 complex plays an important role in the control of melanin production and melanosome biogenesis and promotes the membrane localization of RAB32 and RAB38 (PubMed:23084991).
Tissue Location	Ubiquitous.