Hermansky-Pudlak 综合征蛋白 1 抗体

产品货号： mlR17379

英文名称： HPS1

中文名称： Hermansky-Pudlak 综合征蛋白 1 抗体

别　　名： Hermansky Pudlak syndrome 1 protein; Hermansky Pudlak syndrome gene; Hermansky Pudlak syndrome type 1; Hermansky-Pudlak syndrome 1; Hermansky-Pudlak syndrome 1 protein; HPS; HPS1; HPS1_HUMAN; MGC5277.

研究领域： 细胞生物　信号转导　转运蛋白

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Cow, Horse,

产品应用： ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500 （石蜡切片需

做抗原修复）

not yet tested in other applications.

optimal dilutions/concentrations should be determined by the end user.

分 子 量： 79kDa

细胞定位： 细胞浆 细胞膜

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human HPS1:501-600/700

亚　　型： IgG

纯化方法： affinity purified by Protein A

储 存 液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： Store at -20 ° C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20° C. 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.

PubMed : PubMed

产品介绍 : 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]

Function:

Component of multiple cytoplasmic organelles. Apparently crucial for their normal development and function. May be involved in intracellular protein sorting.

Tissue Specificity:

Ubiquitous.

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.

SWISS:

Q92902

Gene ID:

3257

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

Hermansky－Pudlak 综合征（简称 HPS），是常染色体隐性遗传病，可导致出血时间延长、白化病、溶酶体胶质样沉积等病状。患者通常于 30～50 岁之间死于肺纤维化、出血、结肠炎等严重并发症。目前对该病仍缺乏有效的治疗办法。通过对人、小鼠、酵母等的研究发现，这是一种单基因病，但涉及到多个不同基因的突变。基因突变后，转运途径受阻，表现为黑色素体、溶酶体、血小板致密体等多种亚细胞器的生物合成或功能同时受累的病理改变。对这些 HPS 基因的克隆既有利于进一步阐明发病机制，也有利于建立一系列基因诊断和产前诊断方法，为将来的 HPS 基因治疗提供理论依据。