abcam

Product datasheet

Recombinant Human HPS2 protein ab127282

Description

Product name	Recombinant Human HPS2 protein
Purity	> 95 % SDS-PAGE. Purified via His tag
Expression system	Escherichia coli
Accession	O00203
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Predicted molecular weight	16 kDa
Amino acids	944 to 1094
Tags	His-DHFR tag N-Terminus

Specifications

Our **Abpromise guarantee** covers the use of **ab127282** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	SDS-PAGE
Form	Lyophilized

Preparation and Storage

Stability and Storage	Shipped at 4°C. Store at -20ºC. Constituents: 0.32% Tris HCl, 0.58% Sodium chloride
Reconstitution	Reconstitute with water to desired concentration.

General Info

Function	Subunit of non-clathrin- and clathrin-associated adaptor protein complex 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.
Tissue specificity	Ubiquitously expressed.
Involvement in 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.
Sequence similarities	Belongs to the adaptor complexes large subunit family.
Post-translational modifications	Phosphorylated on serine residues.
Cellular localization	Golgi apparatus. Cytoplasmic vesicle > clathrin-coated vesicle membrane. Golgi apparatus. Component of the coat surrounding the cytoplasmic face of coated vesicles located at the Golgi complex.