abcam

Product datasheet

Recombinant Human Dysbindin protein ab87462

1 Image

Description

Product name	Recombinant Human Dysbindin protein
Purity	> 95 % SDS-PAGE. Purified using conventional chromatography.
Expression system	Escherichia coli
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MRGSHHHHHHGMASMTGGQQMGRDLYDDDDKDRWGS MLSAHWEKKKTSLV ELQEQLQQLPALIADLESMTANLTHLEASFEEVENNLLHLE DLCGQCELE RCKHMQSQQLENYKKNKRKELETFKAELDAEHAQKVLE MEHTQQMKLKER QKFFEEAFQQDMEQYLSTGYLQIAERREPIGSMSSMEVNV DMLEQMDLMD ISDQEALDVFLNSGGEENTVLSPALGPESSTCQNEITLQV PNPSELRAKP PSSSSTCTDSATRDISEGGESPVVQSDEEEVQVDTALAT SHTDREATPDG GEDSDS
Amino acids	1 to 270

Specifications

Our **Abpromise guarantee** covers the use of **ab87462** 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	Liquid

Preparation and Storage

Stability and Storage	Shipped at 4°C. Store at +4°C short term (1-2 weeks). Upon delivery aliquot. Store at -20°C or -

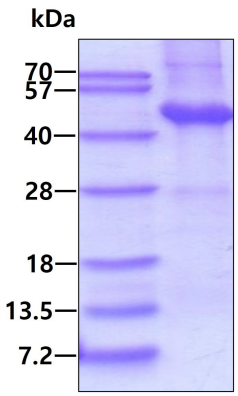
80°C. Avoid freeze / thaw cycle.

pH: 8.00
Constituents: 0.0077% DTT, 0.316% Tris HCl, 20% Glycerol (glycerin, glycerine), 0.58% Sodium chloride

General Info

Function	The BLOC-1 complex is required for normal biogenesis of lysosome-related organelles, such as platelet dense granules and melanosomes. Plays a role in intracellular vesicle trafficking. Plays a role in synaptic vesicle trafficking and in neurotransmitter release. May be required for normal dopamine homeostasis in the cerebral cortex, hippocampus, and hypothalamus. Plays a role in the regulation of cell surface exposure of DRD2. Contributes to the regulation of dopamine signaling. May play a role in actin cytoskeleton reorganization and neurite outgrowth. May modulate MAPK8 phosphorylation.
Tissue specificity	Detected in brain, in neurons and in neuropil. Detected in dentate gyrus and in pyramidal cells of hippocampus CA2 and CA3 (at protein level).
Involvement in disease	Defects in DTNBP1 are the cause of Hermansky-Pudlak syndrome type 7 (HPS7) [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.
Sequence similarities	Belongs to the dysbindin family.
Post-translational modifications	Ubiquitinated by TRIM32. Ubiquitination leads to DTNBP1 degradation. Phosphorylated by PRKDC.
Cellular localization	Cytoplasm. Cytoplasmic vesicle membrane. Cytoplasmic vesicle > secretory vesicle > synaptic vesicle membrane. Endosome membrane. Melanosome membrane. Nucleus. Cell junction > synapse > postsynaptic cell membrane > postsynaptic density. Endoplasmic reticulum. Detected in neuron cell bodies, axons and dendrites. Detected at synapses, at post-synaptic density, at pre-synaptic vesicle membranes and microtubules. Detected at tubulovesicular elements in the vicinity of the Golgi apparatus and of melanosomes. Occasionally detected at the membrane of pigmented melanosomes in cultured melanoma cells.

Images



SDS-PAGE - Recombinant Human Dysbindin protein (ab87462)

3ug by SDS-PAGE under reducing condition and visualized by coomassie blue stain.

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