

## Product datasheet

# Human Dysbindin peptide ab50456

### 概述

产品名称 人Dysbindin肽

### 描述

性质 Synthetic

### 氨基酸序列

种属 Human

序列 C-KTLSDKSREAKVK

氨基酸 21 to 33

### 技术指标

Our [Abpromise guarantee](#) covers the use of **ab50456** in the following tested applications.

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

应用 Blocking

形式 Liquid

### 制备和贮存

稳定性和存储 Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles.

### 常规信息

**功能** 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.

**组织特异性** Detected in brain, in neurons and in neuropil. Detected in dentate gyrus and in pyramidal cells of hippocampus CA2 and CA3 (at protein level).

<b>疾病相关</b>	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.
<b>序列相似性</b>	Belongs to the dysbindin family.
<b>翻译后修饰</b>	Ubiquitinated by TRIM32. Ubiquitination leads to DTNBP1 degradation. Phosphorylated by PRKDC.
<b>细胞定位</b>	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.

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