

## Product datasheet

# Human Dysbindin peptide ab50456

### 製品の概要

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**製品名** Human Dysbindin peptide

### 製品の詳細

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**由来** Synthetic

#### アミノ酸配列

**生物種** Human

**配列** C-KTLSDKSREAKVK

**領域** 21 to 33

### 特性

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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.

**アプリケーション** Blocking

**製品の状態** Liquid

### 前処理および保存

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**保存方法および安定性** Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles.

### 関連情報

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**機能** 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.

**組織特異性** Detected in brain, in neurons and in neuropil. Detected in dentate gyrus and in pyramidal cells of hippocampus CA2 and CA3 (at protein level).

**関連疾患**

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.

**配列類似性**

Belongs to the dysbindin family.

**翻訳後修飾**

Ubiquitinated by TRIM32. Ubiquitination leads to DTNBP1 degradation.  
Phosphorylated by PRKDC.

**細胞内局在**

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.

**Please note:** All products are "FOR RESEARCH USE ONLY AND ARE NOT INTENDED FOR DIAGNOSTIC OR THERAPEUTIC USE"

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