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DTNBP1 Rabbit pAb

Catalog # AP99703

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q96EV8
Other Accession	Q96EV8
Reactivity	Rat
Predicted	Pig, Human, Mouse, Chicken, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	39493
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human DTNBP1
Epitope Specificity	101-200/351
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the dysbindin family.
Post-translational modifications	Ubiquitinated by TRIM32. Ubiquitination leads to DTNBP1 degradation. Phosphorylated by PRKDC.
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.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Dysbindin may play a role in organelle biogenesis associated with melanosomes, platelet dense granules, and lysosomes. A similar protein in mouse is a component of a protein complex termed biogenesis of lysosome-related organelles complex 1 (BLOC1), and binds to alpha and beta dystrobrevins, which are components of the dystrophin associated protein complex (DPC). Mutations in Dysbindin gene are associated with Hermansky-Pudlak syndrome type 7. Dysbindin gene may also be associated with schizophrenia. Multiple transcript variants encoding distinct isoforms have been identified for this gene.

Additional Information

Gene ID	84062
Other Names	DTBP1_HUMAN; DTNBP1; Biogenesis of lysosome-related organelles complex

	1 subunit 8 (BLOC-1 subunit 8); Dysbindin-1; Dystrobrevin-binding protein 1; Hermansky-Pudlak syndrome 7 protein (HPS7 protein); BLOC1S8; DTBP1_MOUSE; Hermansky-Pudlak syndrome 7 protein homolog (HPS7 protein homolog); Sdy; DTBP1_RAT; dystrobrevin binding protein 1; biogenesis of lysosomal organelles complex-1, subunit 8
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

Name	DTNBP1
Synonyms	BLOC1S8
Function	Component of the BLOC-1 complex, a complex that is required for normal biogenesis of lysosome-related organelles (LRO), such as platelet dense granules and melanosomes. In concert with the AP-3 complex, the BLOC-1 complex is required to target membrane protein cargos into vesicles assembled at cell bodies for delivery into neurites and nerve terminals. The BLOC-1 complex, in association with SNARE proteins, is also proposed to be involved in neurite extension. Associates with the BLOC-2 complex to facilitate the transport of TYRP1 independent of AP-3 function. Plays a role in synaptic vesicle trafficking and in neurotransmitter release. Plays a role in the regulation of cell surface exposure of DRD2. May play a role in actin cytoskeleton reorganization and neurite outgrowth. May modulate MAPK8 phosphorylation. Appears to promote neuronal transmission and viability through regulating the expression of SNAP25 and SYN1, modulating PI3-kinase-Akt signaling and influencing glutamatergic release. Regulates the expression of SYN1 through binding to its promoter. Modulates prefrontal cortical activity via the dopamine/D2 pathway.
Cellular Location	[Isoform 1]: Cytoplasm. Cytoplasmic vesicle membrane; Peripheral membrane protein; Cytoplasmic side. Endosome membrane; Peripheral membrane protein; Cytoplasmic side. Melanosome membrane; Peripheral membrane protein; Cytoplasmic side. Postsynaptic density. Endoplasmic reticulum Nucleus. Note=Mainly cytoplasmic but shuttles between the cytoplasm and nucleus. Exported out of the nucleus via its NES in a XPO1-dependent manner. Nuclear localization is required for regulation of the expression of genes such as SYN1 Detected in neuron cell bodies, axons and dendrites. Mainly located to the postsynaptic density. 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. The BLOC-1 complex associates with the BLOC-2 complex in early endosome-associated tubules [Isoform 3]: Cytoplasm. Cytoplasmic vesicle membrane; Peripheral membrane protein; Cytoplasmic side. Cytoplasmic vesicle, secretory vesicle, synaptic vesicle membrane; Peripheral membrane protein; Cytoplasmic side. Endosome membrane; Peripheral membrane protein; Cytoplasmic side. Melanosome membrane; Peripheral membrane protein; Cytoplasmic side. Postsynaptic cell membrane. Endoplasmic reticulum Note=Exclusivley cytoplasmic. Predominantly found in the postsynaptic density (PSD). Little association with synaptic vesicles. The BLOC-1 complex associates with the BLOC-2 complex in early endosome-associated tubules. Associated with the AP-3 complex at presynaptic terminals

Tissue Location	Detected in brain, in neurons and in neuropil. Isoform 1 is expressed in the cerebral cortex, and hippocampal frontal (HF). Specific expression in the posterior half of the superior temporal gyrus (pSTG). Higher expression of isoform 2 and 3 in the HF than in the pSTG while isoform 1 shows no difference in expression in these areas. In the HF, detected in dentate gyrus (DG) and in pyramidal cells of hippocampus CA2 and CA3 (at protein level). Expressed in all principal neuronal populations of the HF, namely pyramidal neurons in the subiculum and CA1-3, granule cells in the dense cell layer of the DG (DGg), and polymorph cells in the hilus of the DG (DGh). Maximal levels in CA2, CA3, and DGh. Isoform 2 not expressed in the cerebral cortex.

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