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PDZD7 Rabbit pAb

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Catalog # AP59117

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	Q9H5P4
Predicted	Rat, Pig, Dog, Human, Mouse, Chicken, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	111752
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human PDZD7
Epitope Specificity	121-220/517
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Cell projection; cilium. Nucleus.
DISEASE	Note=A chromosomal aberration disrupting PDZD7 has been found in patients with non-syndromic sensorineural deafness. Translocation t(10;11),t(10;11). Defects in PDZD7 are a cause of Usher syndrome type 2C (USH2C) [MIM:605472]. USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1), Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH2 is characterized by congenital mild hearing impairment with normal vestibular responses. Note=PDZD7 mutations have been found in combination with mutations in USH2A and GPR98 in patients affected by Usher syndrome, suggesting a role as contributor to digenic Usher syndrome or a modifier of retinal disease expression.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	PDZK7, also known as PDZD7, is a 517 amino acid protein that contains two PDZ (DHR) domains. Encoded by a gene that maps to human chromosome 10q24.31, PDZK7 is conserved in dog, mouse and rat, and exists as three alternatively spliced isoforms. PDZK7 is known to interact with Harmonin, MASS1, USH1G and Usherin. Localizing to nucleus, PDZK7 is expressed in retinal pigment epithelium and inner ear. Biallelic inactivation of PDZK7 can cause non-syndromic hearing impairment and chromosomal aberrations, which are linked to non-syndromic sensorineural deafness. PDZK7 mutations are also linked to Usher syndrome, which is characterized by retinitis pigmentosa and sensorineural deafness, and Alzheimer disease. The gene that encodes PDZK7 maps to human chromosome 10q24.31.

Additional Information

Gene ID	79955
Other Names	PDZ domain-containing protein 7, PDZD7 (HGNC:26257)
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	PDZD7 (HGNC:26257)
Function	In cochlear developing hair cells, essential in organizing the USH2 complex at stereocilia ankle links. Blocks inhibition of adenylate cyclase activity mediated by ADGRV1.
Cellular Location	Cell projection, cilium. Nucleus Cell projection, stereocilium {ECO:0000250|UniProtKB:E9Q9W7} Note=Localizes at the ankle region of the stereocilia {ECO:0000250|UniProtKB:E9Q9W7}
Tissue Location	Weakly expressed in the inner ear. Expressed in the retinal pigment epithelium.

Background

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Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.