



### **PDZD7 Polyclonal Antibody**

Purified Rabbit Polyclonal Antibody (Pab) Catalog # AP59117

### **Specification**

### **PDZD7 Polyclonal Antibody - Product Information**

Application
Primary Accession
Reactivity
Host
Clonality
Calculated MW
Physical State
Immunogen

Epitope Specificity Isotype **Purity** affinity purified by Protein A

Buffer

SUBCELLULAR LOCATION SIMILARITY SUBUNIT

**DISEASE** 

Important Note

WB, IHC-P, IHC-F, IF, E

<u>09H5P4</u>
Rat, Pig, Dog, Bovine
Rabbit
Polyclonal
56 KDa
Liquid
KLH conjugated synthetic peptide derived
from human PDZD7
121-220/517

laG

0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol. Cell projection; cilium. Nucleus. Contains 2 PDZ (DHR) domains. Interacts with USH1G. Interacts with GPR98. Interacts with USH2A. 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. 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.

### PDZD7 Polyclonal Antibody - Additional Information

#### **Gene ID 79955**

#### **Other Names**

PDZ domain-containing protein 7, PDZD7 (<a href="http://www.genenames.org/cgi-bin/gene\_symbol\_report?hgnc\_id=26257" target=" blank">HGNC:26257</a>)

### Target/Specificity

Weakly expressed in the inner ear. Expressed in the retinal pigment epithelium.

### **Dilution**

```
<span class ="dilution_WB">WB~~1:1000</span><br \><span class
="dilution_IHC-P">IHC-P~~N/A</span><br \><span class
="dilution_IHC-F">IHC-F~~N/A</span><br \><span class
="dilution_IF">IF~~1:50~200</span><br \><span class ="dilution_E">E~~N/A</span>
```

### **Format**

0.01M TBS(pH7.4), 0.09% (W/V) sodium azide and 50% Glyce

### **Storage**

Store at -20  $^{\circ}$ 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  $^{\circ}$ C.

### **PDZD7 Polyclonal Antibody - 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.



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# **PDZD7 Polyclonal Antibody - Protocols**

Provided below are standard protocols that you may find useful for product applications.

- Western Blot
- Blocking Peptides
- Dot Blot
- <u>Immunohistochemistry</u>
- <u>Immunofluorescence</u>
- <u>Immunoprecipitation</u>
- Flow Cytomety
- Cell Culture

PDZD7 Polyclonal Antibody - Images