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Rabbit Anti-PDZD7 antibody

SL9042R

Product Name:	PDZD7
Chinese Name:	PDZ结构域PDZK7蛋白抗体
Alias:	PDZ domain containing 7; PDZK7; RP11-108L7.9; EG435601; OTTMUSP00000044305; 9130207N01; OTTMUSP00000044304; PDZD7_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Dog,Pig,Cow,Sheep,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=1:50-200（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	56kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human PDZD7:121-220/517
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. 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.
PubMed:	PubMed
Product Detail:	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.

Subunit:
Interacts with USH1G. Interacts with GPR98. Interacts with USH2A.

Subcellular Location:
Cell projection; cilium. Nucleus.

Tissue Specificity:
Weakly expressed in the inner ear. Expressed in the retinal pigment epithelium.

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.

Similarity:
Contains 2 PDZ (DHR) domains.

SWISS:
Q9H5P4

Gene ID:
79955

Database links:

Entrez Gene: 79955 Human

Omim: 612971 Human

SwissProt: Q9H5P4 Human

Unigene: 438245 Human

	Important Note: This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.