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DFNB31 Rabbit pAb

Catalog # AP98933

Product Information

Application	IHC-P, IHC-F, IF, E
Primary Accession	Q9P202
Other Accession	Q9P202
Predicted	Rat, Human, Mouse, Rabbit, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	96558
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human DFNB31
Epitope Specificity	151-250/907
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Contains 3 PDZ (DHR) domains.
DISEASE	Defects in WHRN are the cause of deafness autosomal recessive type 31 (DFNB31) [MIM:607084]. DFNB31 is a form of sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. Defects in WHRN are the cause of Usher syndrome type 2D (USH2D) [MIM:611383]. USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa and 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.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene is thought to function in the organization and stabilization of sterocilia elongation and actin cystoskeletal assembly, based on studies of the related mouse gene. Mutations in this gene have been associated with autosomal recessive non-syndromic deafness and Usher Syndrome. Alternative splicing of this gene results in multiple transcript variants encoding different isoforms.[provided by RefSeq, Mar 2010]

Additional Information

Gene ID	25861
Other Names	CIP98; DFNB31; PDZD7B; USH2D; WI; 1110035G07Rik; C430046P22Rik; WHRN_HUMAN; WHRN; Autosomal recessive deafness type 31 protein; KIAA1526; WHRN_MOUSE; WHRN_RAT; CASK-interacting protein CIP98; whirlin; deafness, autosomal recessive 31

Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=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	WHRN (HGNC:16361)
Function	Involved in hearing and vision as member of the USH2 complex. Necessary for elongation and maintenance of inner and outer hair cell stereocilia in the organ of Corti in the inner ear. Involved in the maintenance of the hair bundle ankle region, which connects stereocilia in cochlear hair cells of the inner ear. In retina photoreceptors, required for the maintenance of periciliary membrane complex that seems to play a role in regulating intracellular protein transport.
Cellular Location	Cytoplasm {ECO:0000250|UniProtKB:Q80VW5}. Cell projection, stereocilium {ECO:0000250|UniProtKB:Q80VW5}. Cell projection, growth cone {ECO:0000250|UniProtKB:Q80VW5}. Photoreceptor inner segment {ECO:0000250|UniProtKB:Q80VW5}. Synapse {ECO:0000250|UniProtKB:Q810W9}. Note=Detected at the level of stereocilia in inner and outer hair cells of the cochlea and vestibule Localizes to both tip and ankle-link stereocilia regions. Colocalizes with the growing ends of actin filaments. Colocalizes with MPP1 in the retina, at the outer limiting membrane (OLM), outer plexifirm layer (OPL), basal bodies and at the connecting cilium (CC). In photoreceptors, localizes at a plasma membrane microdomain in the apical inner segment that surrounds the connecting cilia called periciliary membrane complex. {ECO:0000250|UniProtKB:Q80VW5, ECO:0000250|UniProtKB:Q810W9, ECO:0000269|PubMed:17584769}

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