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DFNB31 Antibody (Center)

Affinity Purified Rabbit Polyclonal Antibody (Pab)
Catalog # AP11113c

Product Information

Application	IHC-P, WB, E
Primary Accession	Q9P202
Other Accession	NP_056219.3, NP_001077354.2
Reactivity	Human
Host	Rabbit
Clonality	Polyclonal
Isotype	Rabbit IgG
Clone Names	RB19059
Calculated MW	96558
Antigen Region	378-406

Additional Information

Gene ID	25861
Other Names	Whirlin, Autosomal recessive deafness type 31 protein, DFNB31, KIAA1526, WHRN
Target/Specificity	This DFNB31 antibody is generated from rabbits immunized with a KLH conjugated synthetic peptide between 378-406 amino acids from the Central region of human DFNB31.
Dilution	IHC-P~~1:100~500 WB~~1:1000 E~~Use at an assay dependent concentration.
Format	Purified polyclonal antibody supplied in PBS with 0.09% (W/V) sodium azide. This antibody is purified through a protein A column, followed by peptide affinity purification.
Storage	Maintain refrigerated at 2-8°C for up to 2 weeks. For long term storage store at -20°C in small aliquots to prevent freeze-thaw cycles.
Precautions	DFNB31 Antibody (Center) is for research use only and not for use in diagnostic or therapeutic procedures.

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}

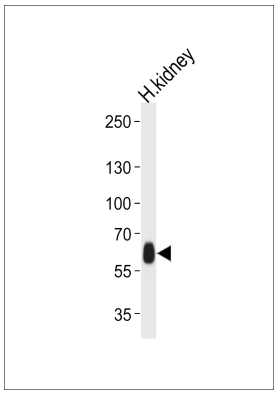
Background

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.

References

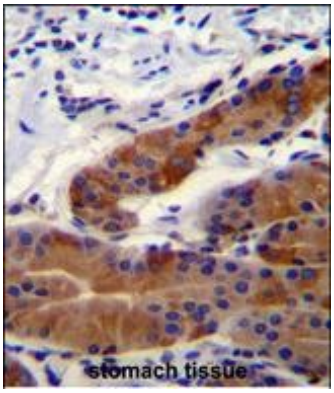
Letra, A., et al. Am. J. Med. Genet. A 152A (7), 1701-1710 (2010) :
Secolin, R., et al. Psychiatr. Genet. 20(3):126-129(2010)
Aller, E., et al. Mol. Vis. 16, 495-500 (2010) :
Toiyama, Y., et al. Int. J. Oncol. 35(4):709-715(2009)
Gosens, I., et al. Hum. Mol. Genet. 16(16):1993-2003(2007)

Images



Western blot analysis of lysate from human kidney tissue lysate, using DFNB31 Antibody (Center)(Cat. #AP11113c). AP11113c was diluted at 1:1000 at each lane. A goat anti-rabbit IgG H&L(HRP) at 1:5000 dilution was used as the secondary antibody. Lysate at 35ug per lane.

DFNB31 Antibody (Center) (Cat. #AP11113c)immunohistochemistry analysis in formalin fixed and paraffin embedded stomach tissue followed by peroxidase conjugation of the secondary antibody and DAB staining.This data demonstrates the use of DFNB31 Antibody (Center) for immunohistochemistry. Clinical relevance has not been evaluated.



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