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Rabbit Anti-DFNB31 antibody

SL14288R

Product Name	DFNB31
Chinese Name	常染色体隐性遗传性耳聋型 31 蛋白抗体
Alias	1110035G07Rik; Autosomal recessive deafness type 31 protein; CASK interacting protein CIP98; CIP 98; CIP98; Deafness autosomal recessive 31; DFNB 31; DKFZp434N014; KIAA1526; RP11 9M16.1; USH 2D; USH2D; Whirlin; WHRN; WHRN_HUMAN; WI.
Research Area	Cell biology immunology Neurobiology
Immunogen Species	Rabbit
Clonality	Polyclonal
React Species	(predicted: Human, Mouse, Rat, Cow, Horse, Rabbit, Sheep,)
Applications	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000 （Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Theoretical molecular weight	96kDa
Cellular localization	The nucleus cytoplasmic
Form	Liquid
Concentration	1mg/ml
immunogen	KLH conjugated synthetic peptide derived from human DFNB31: 151-250/907
Lsotype	IgG
Purification	affinity purified by Protein A
Buffer Solution	1M TBS(pH7.4) with 1% BSA, 3% Proclin300 and 50% Glycerol.
Storage	Shipped at 4℃. Store at -20 °C for one year. Avoid repeated freeze/thaw cycles.
Attention	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
PubMed	PubMed

Product Detail

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]

Function:
Necessary for elongation and maintenance of inner and outer hair cell stereocilia in the organ of Corti in the inner ear.

Subcellular Location:
Cytoplasm. Cell projection > stereocilium. Cell projection > growth cone. Detected at the level of stereocilia in inner outer hair cells of the cochlea and vestibule. Co-localizes with the growing ends of actin filaments (By similarity). Colocalizes with MPP1 in the retina, at the outer limiting membrane (OLM), outer plexifirm layer (OPL), basal bodies and at the connecting cilium.

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.

Similarity:
Contains 3 PDZ (DHR) domains.

SWISS:
Q9P202

Gene ID:
25861

Database links:

Entrez Gene: 25861 Human

Entrez Gene: 73750 Mouse

Entrez Gene: 313255 Rat

Omim: 607928 Human

SwissProt: Q9P202 Human

SwissProt: Q80VW5 Mouse

SwissProt: Q810W9 Rat

Unigene: 93836 Human

Unigene: 300397 Mouse

Unigene: 204268 Rat