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Rabbit Anti-LOXHD1 antibody

SL18343R

Product Name:	LOXHD1
Chinese Name:	脂氧合酶同源结构域1抗体
Alias:	DFNB77; FLJ32670; LH2D1; Lipoxygenase homology domain-containing protein 1; Lipoxygenase homology domains 1; LOXH1_HUMAN; LOXHD1.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Pig,Cow,Horse,Sheep,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	222kDa
Cellular localization:	The cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human LOXHD1:1171-1270/1947
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:	This gene encodes a highly conserved protein consisting entirely of PLAT (polycystin/lipoxygenase/alpha-toxin) domains, thought to be involved in targeting proteins to the plasma membrane. Studies in mice show that this gene is expressed in the mechanosensory hair cells in the inner ear, and mutations in this gene lead to auditory defects, indicating that this gene is essential for normal hair cell function. Screening of human families segregating deafness identified a mutation in this gene

which causes DFNB77, a progressive form of autosomal-recessive nonsyndromic hearing loss (ARNSHL). Alternatively spliced transcript variants encoding different isoforms have been noted for this gene. [provided by RefSeq, Mar 2010]

Function:
Involved in hearing. Required for normal function of hair cells in the inner ear.

DISEASE:
Defects in LOXHD1 are the cause of deafness autosomal recessive type 77 (DFNB77) [MIM:613079]. A form of non-syndromic deafness characterized by preserved low-frequency hearing, and a trend toward mild to moderate mid-frequency and high-frequency hearing loss during childhood and adolescence. Hearing loss progresses to become moderate to severe at mid and high frequencies during adulthood.

Similarity:
Contains 14 PLAT domains.

SWISS:
Q8IVV2

Gene ID:
125336

Database links:

Entrez Gene: 125336 Human

Omim: 613072 Human

SwissProt: Q8IVV2 Human

Unigene: 345877 Human

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