

Recombinant Human LOXHD1 Protein, GST-tagged

Cat. No. LOXHD1-4733H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human LOXHD1 full-length ORF (AAH47720.1, 1 a.a. - 512 a.a.) recombinant protein with GST-tag at N-terminal.
Species	Human
Source	Wheat Germ
Description	This gene encodes a highly conserved protein consisting entirely of PLAT (polycystin/ lipoygenase/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 mechano sensory 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 DFNB7 7, 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]
Molecular Mass	85.1 kDa
AA Sequence	MTVWTGDVVGGGTDSNIFMTLYGINGSTEEMQLDKKKARFEREQNDTFIMEILDIAP FTKMRIRIDGLSRPEWFLERILLKNMNTGDLTMFYYGDWLSQRKGKKTLCVCEMCA VIDEEEMMEWTSYTVAVKTSILGAGTDANVFIIIFGENGDSGTLALKQSANWNKFER NNTDTFNFPDMLSLGHLCKLRVWHDNKGIFPGWHLSDVDKDNSRDETFHFQCDC WLSKSEGDGQTVRDFACANNKICDELETTYEIVIETGNGGETRENVWLILEGRKNR

 Tel: 1-631-559-9269 1-516-512-3133

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

SKEFLMENSSRQRAFRKGTDTTFEFDISIYLGDIASLCVGHlareDRFIPKRELVWHV
 KTITITEMEYGNVYFFNCDCCLIPLKRKRKYFKVFEVTKTTESFASKVQSLVPVKYEVIV
 TTGYEPGAGTDANVFVTIFGANGDTGKRELKQKMRNLFERGSTDRFFLETLELGEL
 RKVRLEHDSSGYCSGWLVEKVEVTNTSTGVATIFNCGRWLDKKRGDGLTWRDLFP
 SV

Applications

Enzyme-linked Immunoabsorbent Assay
 Western Blot (Recombinant protein)
 Antibody Production
 Protein Array

Notes

Best use within three months from the date of receipt of this protein.

Storage

Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.

Storage Buffer

50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

GENE INFORMATION

Gene Name

[LOXHD1 lipoygenase homology domains 1 \[Homo sapiens \(human\) \]](#)

Official Symbol

[LOXHD1](#)

Synonyms

LOXHD1; lipoygenase homology domains 1; LH2D1; DFNB77; lipoygenase homology domain-containing protein 1

Gene ID

[125336](#)

mRNA Refseq

[NM_001145472](#)

Protein Refseq

[NP_001138944](#)

 Tel: 1-631-559-9269 1-516-512-3133

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

MIM	613072
-----	--------

UniProt ID	Q8IVV2
------------	--------

 Tel: 1-631-559-9269 1-516-512-3133

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA