abcam

Product datasheet

Recombinant Human USH1C/Harmonin protein ab117205

Overview

Product name	Recombinant Human USH1C/Harmonin protein
Protein length	Full length protein

Description

Nature	Recombinant
Source	Escherichia coli
Amino Acid Sequence	
Accession	Q9Y6N9
Species	Human
Sequence	MRGSHHHHHH GMASMTGGQQ MGRDLYDDDD KDRWGSHMDR KVAREFRHKV DFLIENDAEK DYLYDVLRMY HQTMDVAVLV GDLKLVINEP SRLPLFDAIR PLIPLKHQVE YDQLTPRRSR KLKEVRLDRL HPEGLGLSVR GGLEFGCGLF ISHLIKGGQA DSVGLQVGDE IVRINGYSIS SCTHEEVINL IRTKKTVSIK VRHIGLIPVK SSPDEPLTWQ YVDQFVSESG GVRGSLGSPG NRENKEKKVF ISLVGSRGLG CSISSGPIQK PGIFISHVKP GSLSAEVGLE IGDQIVEVNG VDFSNLDHKE GRELFMTDRE RLAEARQREL QRQELLMQKR LAMESNKILQ EQQEMERQRR KEIAQKAAEE NERYRKEMEQ IVEEEEKFKK QWEEDWGSKE QLLLPKTITA EVHPVPLRKP KYDQGVEPEL EPADDLDGGT EEQGEQDFRK YEEGFDPYSM FTPEQIMGKD VRLLRIKKEG SLDLALEGGV DSPIGKVVVS AVYERGAAER HGGIVKGDEI MAINGKIVTD YTLAEADAAL QKAWNQGGDW IDLVVAVCPP KEYDDELTFF
Molecular weight	65 kDa including tags
Amino acids	1 to 533
Tags	His tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab117205** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	SDS-PAGE
Purity	> 95 % SDS-PAGE. ab117205 is purified by proprietary chromatographic techniques. Purity was determined to be greater than 95% as determined by RP-HPLC and SDS-PAGE.
Form	Liquid
Additional notes	USH1C/Harmonin protein although stable 4°C for 4 weeks, should be stored desiccated below -18°C. For long term storage it is recommended to add a carrier protein (0.1% HSA or BSA). Avoid freeze-thaw cycles. This product was previously labelled as USH1C

Preparation and Storage

Stability and Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.32% Tris HCl, 20% Glycerol

General Info

Function	May be involved in protein-protein interaction.
Tissue specificity	Expressed in small intestine, colon, kidney, eye and weakly in pancreas. Expressed also in vestibule of the inner ear.
Involvement in disease	Defects in USH1C are the cause of Usher syndrome type 1C (USH1C) [MIM:276904]; also known as Usher syndrome type I Acadian variety. 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). USH1 is characterized by profound congenital sensorineural deafness, absent vestibular function and prepubertal onset of progressive retinitis pigmentosa leading to blindness. Defects in USH1C are the cause of deafness autosomal recessive type 18 (DFNB18) [MIM:602092]. DFNB18 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.
Sequence similarities	Contains 3 PDZ (DHR) domains.
Domain	The PDZ domain 1 mediates interactions with USH1G/SANS and SLC4A7.

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- Response to your inquiry within 24 hours
- We provide support in Chinese, English, French, German, Japanese and Spanish
- Extensive multi-media technical resources to help you
- We investigate all quality concerns to ensure our products perform to the highest standards

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