9051 Mira Mesa Blvd, #910094
San Diego, CA 92191
Tel: 760-390-3989

USH1C Human

Usher Syndrome 1C Human Recombinant
RCP2525

Product Overview

Name	USH1C Human
Description	
Usher Syndrome 1C Human Recombinant	
Accession (Primary)	Q9Y6N9
Synonyms	
Harmonin, Usher syndrome type-1C protein, Autoimmune enteropathy-related antigen AIE-75, Antigen NY-CO-38/NY-CO-37, PDZ-73 protein, Renal carcinoma antigen NY-REN-3, USH1C, AIE75, PDZ73, AIE-75, DFNB18, PDZ-45, NY-CO-37, NY-CO-38, ush1cpst, PDZ-73/NY-CO-38.	
Introduction	
USH1C gene product Harmonin, is a scaffold protein which functions in the assembly of Usher protein complexes. Harmonin is able to attach to various proteins in cell membranes and coordinate their activities. Harmonin contains PDZ domains, a coiled-coil region with a bipartite nuclear localization signal and a PEST degradation sequence. USH1C is expressed in the small intestine, colon, kidney, eye, vestibule of the inner ear and weakly in the pancreas. Mutations in the USH1C gene cause the Usher syndrome type I which is an autosomal recessive sensory defect involving congenital profound sensorineural deafness, vestibular dysfunction, and blindness due to progressive retinitis pigmentosa. Sensorineural deafness is caused by 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. The 3 types of the Usher syndrome (1- 3) are distinguished by age at onset and differences in auditory and vestibular function. USH1C gene defects cause of non-syndromic sensorineural deafness autosomal recessive type 18 (DFNB18), is a form of sensorineural hearing loss.	
Source	
Escherichia Coli.	
Physical Appearance	
Sterile Filtered colorless solution.	
Formulation	
USH1C protein solution (1mg/ml) containing 20mM Tris-HCl buffer (pH 8.0) and 20% glycerol.	
Stability	
Store at 4°C if entire vial will be used within 2-4 weeks. Store, frozen at -20°C for longer periods of time. For long term storage it is recommended to add a carrier protein (0.1% HSA or BSA). Avoid multiple freeze-thaw cycles.	

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Purity

Greater than 95.0% as determined by SDS-PAGE.

Amino acid sequence

MRGSHHHHHH GMASMTGGQQ MGRDLYDDDD KDRWGSH MDR 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.

Precautions

USH1C Human is for research use only and not for use in diagnostic or therapeutic procedures.

Target Information: (Q9Y6N9)