Recombinant Human USH1C, His-tagged

Cat. No. USH1C-29611TH **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant fragment, corresponding to amino acids 1-533 of human USH1C fused to His tag at N-terminus; 570 amino acids, 64.6 kDa.
Species	Human
Source	E.coli
ProteinLength	1-533 a.a.
Description	This gene encodes a scaffold protein that functions in the assembly of Usher protein complexes. The protein contains PDZ domains, a coiled-coil region with a bipartite nuclear localization signal and a PEST degradation sequence. Defects in this gene are the cause of Usher syndrome type 1C and non-syndromic sensorineural deafness autosomal recessive type 18. Multiple transcript variants encoding different isoforms have been found for this gene.
Conjugation	HIS
Tissue specificity	Expressed in small intestine, colon, kidney, eye and weakly in pancreas. Expressed also in vestibule of the inner ear.
Form	Liquid
Purity	>95% by SDS-PAGE

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Storage buffer	Preservative: NoneConstituents: 20% Glycerol, 20mM Tris HCl, pH 8.0
Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.
Sequences of amino acids	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
Sequence Similarities	Contains 3 PDZ (DHR) domains.

GENE INFORMATION

Gene Name	USH1C Usher syndrome 1C (autosomal recessive, severe) [Homo sapiens]
Official Symbol	USH1C
Synonyms	USH1C; Usher syndrome 1C (autosomal recessive, severe); deafness, autosomal recessive 18 , DFNB18; harmonin; AIE 75; NY CO 37; NY CO 38; PDZ 73; PDZ73;
Gene ID	10083

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mRNA Refseq	NM_005709
Protein Refseq	NP_005700
MIM	605242
Uniprot ID	Q9Y6N9
Chromosome Location	11p14.3
Function	protein binding;

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