Recombinant Human USH1C, His-tagged

Cat. No. USH1C-29610TH **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant full length, Human USH1C (amino acids 1-533) with N terminal His tag; 570aa, 64.6kDa.
Species	Human
Source	E.coli
ProteinLength	533 amino acids
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
Molecular Weight	64.600kDa inclusive of tags
Tissue specificity	Expressed in small intestine, colon, kidney, eye and weakly in pancreas. Expressed also in vestibule of the inner ear.
Form	Liquid

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Purity	>95% by SDS-PAGE
Storage buffer	pH: 8.00Constituents:0.32% Tris HCl, 20% Glycerol
Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid 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;

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Gene ID	10083
mRNA Refseq	NM_005709
Protein Refseq	NP_005700
MIM	605242
Uniprot ID	Q9Y6N9
Chromosome Location	11p14.3
Function	protein binding;

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