

Recombinant Human Usher Syndrome 1C, His-tagged

Cat. No. USH1C-475H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant human Harmonin protein, fused to His-tag at N-terminus, was expressed in <i>E.coli</i> and purified by using conventional chromatography. MW = 64.6 kDa (570aa).
Species	Human
Source	E.coli
Description	Harmonin, product encoded by USH1c gene, is a scaffold protein that functions in the assembly of Usher protein complexes. Usher syndrome type I caused by mutations in USH1C is an autosomal recessive sensory defect involving congenital profound sensorineural deafness, vestibular dysfunction, and blindness due to progressive retinitis pigmentosa. Harmonin has the ability to bind to many other proteins in cell membranes and coordinates their activities.
Amino Acid Sequence	MRGSHHHHHH GMASMTGGQQ MGRDLYDDDD KDRWGSHMDR KVAREFRHKV DFLIENDA EK DYLYDVLRMY HQTMDVAVLV GDLKLVINEP SRLPLFDAIR PLIPLKHQVE YDQLTPRRSR KLKEVRLDRL HPEGLGLSVR GGLEFGCGLF ISHLIKGGQA DSVGLQVGDE IVRINGYSIS SCTHEEVINL IRTKKT VSIK VRHIGLIPVK SSPDEPLTWQ YVDQFVSESG GVRGSLGSPG NRENKEKKVF ISLVGSRGLG CSISSGPIQK PGIFISHVKP GSLSAEVLG IGDQIVEVNG VDFSNL DHKE GRELFMTDRE RLAEARQREL QRQELLMQKR LAMESNKILQ EQQEMERQRR KEIAQKAAEE NERYRKEMEQ IVEEEEKFKK QWEEDWGSKE QLLLPKTITA EVHPVPLRKP KYDQGVEPEL EPADDLDGGT EEQGEQDFRK

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YEEGFDPYSM FTPEQIMGKD VRLRIKKEG SLDLALEGGV DSPIGKVVVS
 AVYERGAAER HGGIVKGDEI MAINGKIVTD YTLAEADAAL QKAWNQGQGDW
 IDLVVAVCPP KEYDDELTFF.

Form	Liquid. In 20 mM Tris-HCl buffer (pH 8.0) containing 20% glycerol.
Purity	> 95% by SDS – PAGE.
Concentration	1 mg/ml (determined by Bradford assay).
Storage	Can be stored at +4°C short term (1-2 weeks). For long term storage, aliquot and store at -20°C or -70°C. Avoid repeated freezing and thawing cycles.

GENE INFORMATION

Gene Name	USH1C Usher syndrome 1C (autosomal recessive, severe) [Homo sapiens]
Synonyms	USH1C; Usher syndrome 1C (autosomal recessive, severe); PDZ73; AIE-75; DFNB18; PDZ-45; PDZ-73; NY-CO-37; NY-CO-38; ush1cpst; PDZ-73/NY-CO-38; harmonin; deafness, autosomal recessive 18; PDZ73; PDZ-73; 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; AIE75
Gene ID	10083
mRNA Refseq	NM_005709
Protein Refseq	NP_005700
MIM	605242

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UniProt ID	Q9Y6N9
Chromosome Location	11p14.3
Function	protein binding
PDB rendering based on 1v6b	