

## 32-5201: Recombinant Human Usher Syndrome 1C

**Alternative Name :** 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

### Description

Source : Escherichia Coli. USH1C Human Recombinant fused with a 37 amino acid His tag at N-terminus produced in E.Coli is a single, non-glycosylated, polypeptide chain containing 570 amino acids (1-533 a.a.) and having a molecular mass of 64.6kDa.The USH1C is purified by proprietary chromatographic techniques. 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.

### Product Info

**Amount :** 20 µg

**Purification :** Greater than 95.0% as determined by SDS-PAGE.

**Content :** USH1C protein solution (1mg/ml) containing 20mM Tris-HCl buffer (pH 8.0) and 20% glycerol.

**Storage condition :** 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.

**Amino Acid :** MRGSHHHHHH GMASMTGGQQ MGRDLYDDDD KDRWGSHMDR KVAREFRHKV DFLIENDA EK DYLYDVL RMY HQTMDVAVLV GDLKLVINEP SRLPLFDAIR PLIPLKHQVE YDQLTPRRSR KLKEVRLDRL HPEGLGLSVR GGLEFGCGLF ISHLIKGGQA DSVGLQVGDE IVRINGYSIS SCTHEEVINL IRTKKT VSIK VRHIGLIPVK SSPDEPLTWQ YVDQFVSESG GVRGSLGSPG NRENKEKKVF ISLVGSRGLG CSISSGPIQK PGIFISHVKP GLSAEVGLE IGDQIVEVNG VDFSNL DHKE GRELFMTDRE RLAEARQREL QRQELLMQKR LAMESNKILQ EQQEMERQRR KEIAQKAAEE NERYRKEMEQ IVEEEEFKFK QWEEDWGSKE QLLPKTITA EVHPVPLRKP KYDQGVPEL EPADDLDGGT EEQGEQDFRK YEEGFDPSM FTPEQIMGKD VRLRIKKEG SLDLALEGGV DSPIGKVVVS AVYERGA AER HGGIVKGDEI MAINGKIVTD YTLAEADAAL QKAWNQGGDW IDLVVAVCPP KEYDDELTFE.

