

# Recombinant human Harmonin/USH1C protein

Catalog Number: ATGP0337

## PRODUCT INFORMATION

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**Expression system**

E.coli

**Domain**

1-533aa

**UniProt No.**

Q9Y6N9

**NCBI Accession No.**

AAH16057

**Alternative Names**

AIE 75, Autoimmune enteropathy related antigen AIE 75, Deafness autosomal recessive 18, DFNB 18, Harmonin, NY CO 37, NY CO 38, PDZ 45, PDZ 73, Renal carcinoma antigen NY REN 3, USH 1C, Ush1cpst, Usher syndrome 1C (autosomal recessive severe), Usher syndrome type 1C protein

## PRODUCT SPECIFICATION

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**Molecular Weight**

64.6 kDa (570aa)

**Concentration**

1mg/ml (determined by Bradford assay)

**Formulation**

Liquid in. 20mM Tris-HCl buffer (pH 8.0) containing 20% glycerol

**Purity**

&gt; 95% by SDS-PAGE

**Tag**

His-Tag

**Application**

SDS-PAGE

**Storage Condition**

Can be stored at +2C to +8C for 1 week. For long term storage, aliquot and store at -20C to -80C. Avoid repeated freezing and thawing cycles.

## BACKGROUND

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**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

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their activities. Recombinant human Harmonin protein, fused to His-tag at N-terminus, was expressed in *E. coli* and purified by using conventional chromatography.

## Amino acid Sequence

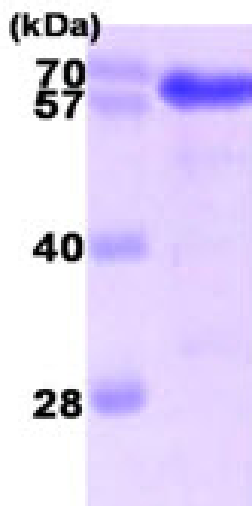
MRGSHHHHHH GMASMTGGQQ MGRDLYDDDD KDRWGSMDR KVAREFRHKV DFLIENDA EK DYLYDVLRMY  
HQTMDEVAVLV GDLKLVINEP SRLPLFDAIR PLIPLKHQVE YDQLTPRRSR KLKEVRLDRL HPEGLGLSVR GGLEFGCGLF  
ISHLIKGGQA DSVGLQVGDE IVRINGYSIS SCTHEEVINL IRTKKT VSIK VRHIGLIPVK SSPDEPLTWQ YVDQFVSESG  
GVRGSLGSPG NRENKEKKVF ISLVGSRGLG CSISSGPIQK PGIFISHVKP GSLSAEVGL E IGDQIVEVNG VDFS NLDHKE  
GRELFMTDRE RLAEARQREL QRQELLMQKR LAMESNKILQ EQQEMERQRR KEIAQKAAEE NERYRKEMEQ IVEEEEEFKK  
QWEEDWGSKE QLLLPKTITA EVHPVPLRKP KYDQGVPEL EPADDLDGGT EEQGEQDFRK YEEGFDPYSM FTPEQIMGKD  
VRLLRKKEG SLDLALEGGV DSPIGKVVVS AVYERGAAER HGGIVKGDEI MAINGKIVTD YTLAEADAAL QKAWNQGGDW  
IDLVVAVCPP KEYDDELTF

## General References

Siemens J., et al. (2002). *Proc Natl Acad Sci U S A.* 99(23):14946-51.  
Ouyang XM., et al. (2002). *Hum Genet.* 111(1):26-30.

## DATA

### SDS-PAGE



3ug by SDS-PAGE under reducing condition and visualized by coomassie blue stain.

15% SDS-PAGE (3ug)