

Harmonin/USH1C Protein, Human, Recombinant (His)

General Information

Synonyms:	PDZD7C;PDZ-73;PDZ73;DFNB18A;PDZ-73/NY-CO-38;DFNB18;NY-CO-37;PDZ-45;NY-CO-38; Usher syndrome 1C (autosomal recessive, severe);AIE-75;ush1cpst
Protein Construction:	A DNA sequence encoding the native human USH1C (Q9Y6N9-1) (Met 1-Phe 552) was expressed, with a polyhistidine tag at the N-terminus. Predicted N terminal: Met
Species:	Human
Expression Host:	E. coli
Accession:	Q9Y6N9-1
Molecular Weight:	63.7 kDa (predicted); 63.7 kDa (reducing conditions)

QC Testing

Biological Activity:	Activity testing is in progress. It is theoretically active, but we cannot guarantee it. If you require protein activity, we recommend choosing the eukaryotic expression version first.
Purity:	≥ 90 % as determined by SDS-PAGE
Endotoxin:	Please contact us for more information.
Formulation:	Supplied as sterile 50 mM Tris, 20% glycerol, pH 7.7.

Preparation and Storage

Reconstitution:

A Certificate of Analysis (CoA) containing reconstitution instructions is included with the products. Please refer to the CoA for detailed information.

Stability & Storage:

It is recommended to store the product under sterile conditions at -20°C to -80°C. Samples are stable for up to 12 months. Please avoid multiple freeze-thaw cycles and store products in aliquots.

Actual storage temperature shall be subject to the COA.

Shipping:

Proteins are shipped with blue ice.

Protein Background

Harmonin, also known as Antigen NY-CO-38 / NY-CO-37, Autoimmune enteropathy-related antigen AIE-75, Protein PDZ-73, Renal carcinoma antigen NY-REN-3, Usher syndrome type-1C protein and USH1C, is a protein that is expressed in small intestine, colon, kidney, eye and weakly in pancreas. USH1C is expressed also in vestibule of the inner ear. USH1C contains 3 PDZ (DHR) domains. USH1C may be involved in protein-protein interaction. Defects in USH1C are the cause of Usher syndrome type 1C (USH1C), also known as Usher syndrome type I Acadian variety. USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa and sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1), Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). Defects in USH1C are

also the cause of deafness autosomal recessive type 18 (DFNB18) which is a form of sensorineural hearing loss. Sensorineural deafness results from 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.

Reference

Verpy, E. et al., 2000, Nat Genet. 26 (1):51-5.

Weil D., et al., 2003, Hum. Mol. Genet. 12:463-471.

Reiners,J. et al., 2005, Hum Mol Genet. 14 (24):3933-43.

Yan,D. et al., 2006, Mol Biol. 357 (3):755-64.

Inhibitor · Natural Compounds · Compound Libraries · Recombinant Proteins

This product is for Research Use Only· Not for Human or Veterinary or Therapeutic Use

Tel:781-999-4286 E_mail:info@targetmol.com Address:34 Washington Street,Wellesley Hills,MA 02481