

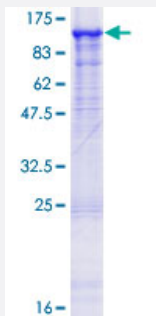
## Full-Length

# OPHN1 (Human) Recombinant Protein (P01)

Catalog # H00004983-P01

Size 25 ug, 10 ug

## Applications



## Specification

### Product Description

Human OPHN1 full-length ORF ( NP\_002538.1, 1 a.a. - 802 a.a.) recombinant protein with GST-tag at N-terminal.

### Sequence

MGHPPLEFSDCYLDSPDFRERLKCYEQELERTNKFIDVIKDGNALISAMRNYSSAVQKFSQTLQ  
SFQDFDFIGDTLTDDEINIAESFKEFAELLNEVENERMMMVMHNASDLLIKPLENFRKEQIGFTKERKK  
KFEKDGERFYSLDRHLHLSSKKKESQLQEADLQVDKERHNFFESSLDYVYQIQEVQESKKFNV  
EPVLAFLHSLFISNSLTVELTQDFLPYKQQLQLSLQNTRNHFSSTREEMEELKKRMKEAPQTCKLP  
GQPTIEGYLTQEKWALGISWVKYYCQYEKETKTLTTPMEQKPGAKQGPLDLTLKYCVRRTESI  
DKRFCFDIETNERPGTITLQALSEANRRLWMEAMDGKEPIYHSPITKQQEMELNEVGKFFVRKCINII  
ETKGIKTEGLYRTVGSNIQVQKLLNAFFDPKCPGDVDFHNSDWDIKTITSSLKFYLRNLSEPVMTYR  
LHKELVSAAKSDNLDYRLGAIHSLVYKLPEKNREMLELLIRHLVNVCEHSKENLMTPSNMGVIFGP  
TLMRAQEDTVAAMMNIKFQNVVEILIEHFGKIYLPPEESAAPPVPPPRVTARRHKPITISKRLLRER  
TVFYTSSLDESEDEIQHQTPNGTITSSIEPPKPPQHPKLPQIRSGETDPGRKSPSRPILDGKLEPCP  
EVDVGKLVSRQLDGGTKITPKATNGPMPGSGPTKTPSFHIKRPAPRPLAHHKEGDADSFASKVRP  
PGEKPTIIRPPVRPPDPPCRAATPQKPEPKPDVAGNAGEITSSVASRTRFFETASRKTGSSQGR  
LPGDES

### Host

Wheat Germ (in vitro)

### Theoretical MW (kDa)

118

### Interspecies Antigen Sequence

Mouse (92); Rat (92)

|                         |                                                                          |
|-------------------------|--------------------------------------------------------------------------|
| Preparation Method      | <a href="#">in vitro wheat germ expression system</a>                    |
| Purification            | Glutathione Sepharose 4 Fast Flow                                        |
| Quality Control Testing | 12.5% SDS-PAGE Stained with Coomassie Blue.                              |
| Storage Buffer          | 50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer. |
| Storage Instruction     | Store at -80°C. Aliquot to avoid repeated freezing and thawing.          |
| Note                    | Best use within three months from the date of receipt of this protein.   |

## Applications

- Enzyme-linked Immunoabsorbent Assay
- Western Blot (Recombinant protein)
- Antibody Production
- Protein Array

## Gene Info — OPHN1

|                     |                                                                                                                                                                                                                                                                                                                                                                                                                   |
|---------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Entrez GeneID       | <a href="#">4983</a>                                                                                                                                                                                                                                                                                                                                                                                              |
| GeneBank Accession# | <a href="#">NM_002547.1</a>                                                                                                                                                                                                                                                                                                                                                                                       |
| Protein Accession#  | <a href="#">NP_002538.1</a>                                                                                                                                                                                                                                                                                                                                                                                       |
| Gene Name           | OPHN1                                                                                                                                                                                                                                                                                                                                                                                                             |
| Gene Alias          | MRX60, OPN1                                                                                                                                                                                                                                                                                                                                                                                                       |
| Gene Description    | oligophrenin 1                                                                                                                                                                                                                                                                                                                                                                                                    |
| Omim ID             | <a href="#">300127</a> <a href="#">300486</a>                                                                                                                                                                                                                                                                                                                                                                     |
| Gene Ontology       | <a href="#">Hyperlink</a>                                                                                                                                                                                                                                                                                                                                                                                         |
| Gene Summary        | This gene encodes a Rho-GTPase-activating protein that promotes GTP hydrolysis of Rho subfamily members. Rho proteins are important mediators of intracellular signal transduction, which affects cell migration and cell morphogenesis. Mutations in this gene are responsible for OPHN1-related X-linked mental retardation with cerebellar hypoplasia and distinctive facial dysmorphism. [ provided by RefSeq |

**Other Designations**

OTTHUMP00000023451|mental retardation, X-linked 60|oligophrenin-1, Rho-GTPase activating protein

**Disease**

- [Angina Pectoris](#)
- [Cerebellar Diseases](#)
- [Coronary Vasospasm](#)
- [Facial Asymmetry](#)
- [Genetic Predisposition to Disease](#)
- [Mental Retardation](#)
- [Nervous System Malformations](#)