# NPC1 Human Recombinant Protein

## PRODUCT OVERVIEW

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| **Product Name** | NPC1, Recombinant Protein |
| **Full Product Name** | Recombinant Human NPC1 |
| **Product Gene Name** | NPC1 recombinant protein |
| **Product Synonym Names** | Niemann Pick C1 protein; Niemann Pick disease; type C1; NPC1 |
| **Host** | E Coli |
| **Form/Format** | Lyophilized from a 0.2um filtered solution in PBS with 5% trehalose, pH7.4 |
| **Purity/Purification** | Greater than 75% by SDS-PAGE gel analyses |
| **Preparation and Storage** | Store at -20 degree C for 12 months as lyophilized; 2-8 degree C for 1 month under sterile conditions after reconstitution |
| **Molecular Weight** | 1278 |
| **Sequence Positions** | 37-261 |
| **OMIM** | phenotype 607623 |
| **3D Structure** | ModBase 3D Structure for O15118 |
| **Species** | Human |
| **Tag** | N-terminal His-IF2DI Tag |
| **Reconstitution** | Centrifuge the vial at 10,000rpm for 1 minute, reconstitute at 200ug/ml in sterile distilled water |
| **ISO Certification** | Manufactured in an ISO 13485:2003 and EN ISO 13485:2012 Certified Laboratory. |
| **Applications Tested/Suitable for NPC1 recombinant protein** | Western Blot (WB), ELISA (EIA) |
| **Related Product Information for NPC1 recombinant protein** | MBS7613520 is a recombinant protein targeting NPC1. Also known as Recombinant Human NPC1. Host: E Coli. |
| **Other Notes** | Small volumes of NPC1 recombinant protein vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply. |

## ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for NPC1. It may not necessarily be applicable to this product.

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| **NCBI GI #** | 83305902 |
| **NCBI GeneID** | 4864 |
| **NCBI Accession #** | O15118.2 |
| **UniProt Primary Accession #** | O15118 |
| **UniProt Secondary Accession #** | Q9P130; B4DET3 |
| **UniProt Related Accession #** | O15118 |
| **NCBI Official Full Name** | Niemann-Pick C1 protein |

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| **NCBI Official Synonym Full Names** | Niemann-Pick disease, type C1 |
| **NCBI Official Symbol** | NPC1 |
| **NCBI Official Synonym Symbols** | NPC |
| **UniProt Protein Name** | Niemann-Pick C1 protein |
| **Protein Family** | Niemann-Pick C1 protein |
| **UniProt Gene Name** | NPC1 |
| **UniProt Entry Name** | NPC1_HUMAN |
| **NCBI Protein Information** | Niemann-Pick C1 protein |
| **NCBI Summary for NPC1** | This gene encodes a large protein that resides in the limiting membrane of endosomes and lysosomes and mediates intracellular cholesterol trafficking via binding of cholesterol to its N-terminal domain. It is predicted to have a cytoplasmic C-terminus, 13 transmembrane domains, and 3 large loops in the lumen of the endosome - the last loop being at the N-terminus. This protein transports low-density lipoproteins to late endosomal/lysosomal compartments where they are hydrolized and released as free cholesterol. Defects in this gene cause Niemann-Pick type C disease, a rare autosomal recessive neurodegenerative disorder characterized by over accumulation of cholesterol and glycosphingolipids in late endosomal/lysosomal compartments.[provided by RefSeq, Aug 2009] |
| **UniProt Comments for NPC1** | NPC1: Intracellular cholesterol transporter which acts in concert with NPC2 and plays an important role in the egress of cholesterol from the endosomal/lysosomal compartment. Both NPC1 and NPC2 function as the cellular 'tag team duo' (TTD) to catalyze the mobilization of cholesterol within the multivesicular environment of the late endosome (LE) to effect egress through the limiting bilayer of the LE. NPC2 binds unesterified cholesterol that has been released from LDLs in the lumen of the late endosomes/lysosomes and transfers it to the cholesterol-binding pocket of the N-terminal domain of NPC1. Cholesterol binds to NPC1 with the hydroxyl group buried in the binding pocket and is exported from the limiting membrane of late endosomes/ lysosomes to the ER and plasma membrane by an unknown mechanism. Binds oxysterol with higher affinity than cholesterol. May play a role in vesicular trafficking in glia, a process that may be crucial for maintaining the structural and functional integrity of nerve terminals. Defects in NPC1 are the cause of Niemann-Pick disease type C1 (NPC1). A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C1 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. An allelic variant of Niemann-Pick disease type C1 is found in people with Nova Scotia ancestry. Patients with the Nova Scotian clinical variant are less severely affected. Belongs to the patched family.<br><br>Protein type: Membrane protein, integral; Membrane protein, multi-pass<br><br>Chromosomal Location of Human Ortholog: 18q11.2<br><br>Cellular Component: Golgi apparatus; membrane; lysosome; perinuclear region of cytoplasm; integral to plasma membrane; lysosomal membrane; late endosome membrane; endoplasmic reticulum; extracellular region; integral to membrane; nuclear envelope; lipid raft<br><br>Molecular Function: protein binding; transmembrane receptor activity; sterol transporter activity; hedgehog receptor activity; cholesterol binding; receptor activity<br><br>Biological Process: response to drug; cholesterol metabolic process; lysosomal transport; cholesterol transport; bile acid metabolic process; protein amino acid glycosylation; cholesterol efflux; endocytosis; signal transduction; adult walking behavior; negative regulation of macroautophagy; cholesterol homeostasis; response to cadmium ion; autophagy; lipid raft organization and biogenesis<br><br>Disease: Niemann-pick Disease, Type C1 |
| **Research Articles on NPC1** | **1.** Data suggest that in order for the ligand cholesterol to slide from one binding pocket to the other (from NPC2 to NPC1), cholesterol undergoes conformational change/isomerization to accommodate the bent transfer pathway between the 2 binding pockets. |

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