

Recombinant Human NPC1

Cat. No. NPC1-29132TH **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant fragment, corresponding to amino acids 151-250 of Human Niemann Pick C1, with an N-terminal proprietary tag, predicted MWt 36.63 kDa
Species	Human
Source	Wheat Germ
ProteinLength	100 amino acids
Description	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 hydrolyzed 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.
Molecular Weight	36.630kDa inclusive of tags
Form	Liquid
Purity	Proprietary Purification

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Storage buffer	pH: 8.00Constituents:0.3% Glutathione, 0.79% Tris HCl
Storage	Shipped on dry ice. Upon delivery aliquot and store at -80oC. Avoid freeze / thaw cycles.
Sequences of amino acids	GFANAMYNACRDVEAPSSNDKALGLLCGKDADACNATNWIEYMFNKDNGQAPFTIT PVFSDFPVHGMEPMNNATKGCDESVDVETAPCSCQDCSIVCGPK
Sequence Similarities	Belongs to the patched family.Contains 1 SSD (sterol-sensing) domain.

GENE INFORMATION

Gene Name	NPC1 Niemann-Pick disease, type C1 [Homo sapiens]
Official Symbol	NPC1
Synonyms	NPC1; Niemann-Pick disease, type C1; Niemann-Pick C1 protein;
Gene ID	4864
mRNA Refseq	NM_000271
Protein Refseq	NP_000262
MIM	607623
Uniprot ID	O15118
Chromosome Location	18q11-q12

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Pathway	Lysosome, organism-specific biosystem; Lysosome, conserved biosystem;
Function	hedgehog receptor activity; protein binding; receptor activity; sterol transporter activity; transmembrane signaling receptor activity;

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