Recombinant Human NPC1, GST-tagged

Cat. No. NPC1-29133TH **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human NPC1(151 a.a. - 250 a.a.), fused with GST-tag at N-terminal, was expressed in wheat germ.
Species	Human
Source	Wheat Germ
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 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.
Molecular Mass	36.63 kDa
AA Sequence	GFANAMYNACRDVEAPSSNDKALGLLCGKDADACNATNWIEYMFNKDNGQAPFTIT PVFSDFPVHGMEPMNNATK GCDESVDEVTAPCSCQDCSIVCGPK
Applications	ELISA; WB-Re; AP; Array

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Storage	Store at -80°C. Aliquot to avoid repeated freezing and thawing.
Storage Buffer	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

GENE INFORMATION

Gene Name	NPC1 Niemann-Pick disease, type C1 [Homo sapiens (human)]
Official Symbol	NPC1
Synonyms	NPC1; NPC; 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	18q11.2
Pathway	Lysosome
Function	cholesterol binding; hedgehog receptor activity; protein binding

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