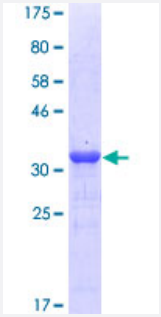
NPC1 (Human) Recombinant Protein (Q01)

Catalog # H00004864-Q01 Size 25 ug, 10 ug

Applications



Specification

Product Description	Human NPC1 partial ORF (AAH63302, 151 a.a. - 250 a.a.) recombinant protein with GST-tag at N-terminal.
Sequence	GFANAMYNACRDVEAPSSNDKALGLLCGKDADACNATNWIEYMFNKDNGQAPFTITPVFSDFPV HGMEPMNNATKGCDESVDEVTAPCSCQDCSIVCGPK
Host	Wheat Germ (in vitro)
Theoretical MW (kDa)	36.63
Interspecies Antigen Sequence	Mouse (89); Rat (89)
Preparation Method	*in vitro* wheat germ expression system
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 — NPC1

Entrez GeneID	4864
GeneBank Accession#	BC063302
Protein Accession#	AAH63302
Gene Name	NPC1
Gene Alias	NPC
Gene Description	Niemann-Pick disease, type C1
Omim ID	257220 607623
Gene Ontology	Hyperlink
Gene Summary	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
Other Designations	-

Pathway

- Lysosome

Disease

- Alzheimer disease
- Birth Weight
- Cardiovascular Diseases
- Coronary Artery Disease
- Coronary Disease
- Diabetes Complications
- Diabetes Mellitus
- Edema
- Genetic Predisposition to Disease
- Lymphoma
- Metabolic Syndrome X
- Neoplasms
- Niemann-Pick Disease
- Obesity
- Osteoporosis
- Overweight