abcam

Product datasheet

Recombinant Human Niemann Pick C1 protein ab114306

1 Image

Overview

Product name	Recombinant Human Niemann Pick C1 protein
Protein length	Protein fragment

Description

Nature	Recombinant
Source	Wheat germ
Amino Acid Sequence	
Accession	O15118
Species	Human
Sequence	GFANAMYNACRDVEAPSSNDKALGLLCGKDADACNATNWIEYMFNKDNGQ APFTITPVFSDFPVHGMEPMNNATKGCDESVDEVTAPCSCQDCSIVCGPK
Molecular weight	37 kDa including tags
Amino acids	151 to 250

Specifications

Our Abpromise guarantee covers the use of **ab114306** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	Western blot SDS-PAGE ELISA
Form	Liquid
Additional notes	Protein concentration is above or equal to 0.05 mg/ml. This protein is best used within three months from the date of receipt.

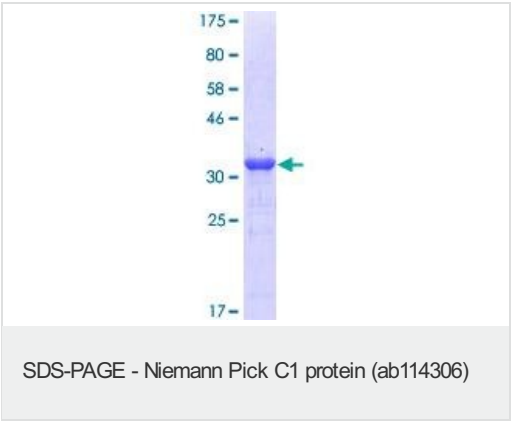
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.3% Glutathione, 0.79% Tris HCl

General Info

Function	Involved in the intracellular trafficking of 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.
Involvement in disease	Defects in NPC1 are the cause of Niemann-Pick disease type C1 (NPDC1) [MIM:257220]. 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.
Sequence similarities	Belongs to the patched family. Contains 1 SSD (sterol-sensing) domain.
Domain	A cysteine-rich N-terminal domain and a C-terminal domain containing a di-leucine motif necessary for lysosomal targeting are critical for mobilization of cholesterol from lysosomes.
Post-translational modifications	Glycosylated.
Cellular localization	Late endosome membrane. Lysosome membrane.

Recombinant Human Niemann Pick C1 protein images



SDS-PAGE - Niemann Pick C1 protein (ab114306)

12.5% SDS-PAGE image showing ab114306
Stained with Coomassie Blue.

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