abcam

Product datasheet

Recombinant Human Niemann Pick C1 protein ab114306

1 Image

Description

Product name	Recombinant Human Niemann Pick C1 protein
Expression system	Wheat germ
Accession	O15118
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	GFANAMYNACRDVEAPSSNDKALGLLCGKDADACNATN WIEYMFNKDNGQ APFTITPVFSDFPVHGMEPMNNATKGCDESVDEVTAPCS CQDCSIVCGPK
Predicted 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

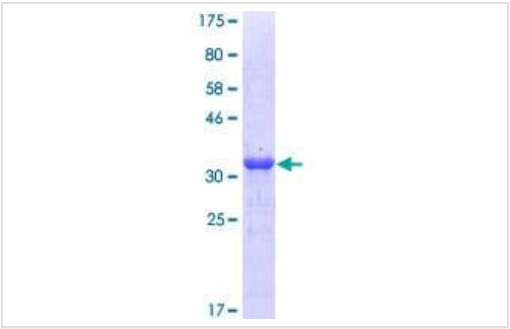
Preparation and Storage

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.

Images



SDS-PAGE - Recombinant Human Niemann Pick C1 protein (ab114306)

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

Please note: All products are "FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC PROCEDURES"

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