SANTA CRUZ BIOTECHNOLOGY, INC.

NPC1 (G-1): sc-271335

SANTA CRUZ BIOTECHNOLOGY

The Power to Question

BACKGROUND

Cells obtain cholesterol via two distinct pathways, endogenous synthesis in the endoplasmic reticulum and exogenous uptake through the low-density lipoprotein (LDL) receptor pathway. NPC1 is a protein that resides in late endosomes and lysosomes and is involved in the intracellular trafficking of cholesterol. The human NPC1 gene maps to chromosome 18q11.2 and produces proteins which undergo N-glycosylation and are expressed in brain and liver. NPC1 contains a cysteine-rich domain, which is critical for proper protein function, but is highly mutated. Mutations in NPC1 result in Niemann-Pick disease type C (NPC), an autosomal recessive disease characterized by the accumulation of unesterified cholesterol in the endosomal/lysosomal system. The accumulation of cholesterol results in progressive neurodegeneration and death. More than 90% of cases of NPC are due to mutations in NPC1 and patients with NPC display multiple neurological symptoms, such as hepatosplenomegaly, ataxia, dystonia and dementia.

REFERENCES

1. Watari, H., et al. 1999. Mutations in the leucine zipper motif and sterol-sensing domain inactivate the Niemann-Pick C1 glycoprotein. J. Biol. Chem. 274: 21861-21866.
2. Greer, W.L., et al. 1999. Mutations in NPC1 highlight a conserved NPC1-specific cysteine-rich domain. Am. J. Hum. Genet. 65: 1252-1260.
3. Sym, M., et al. 2000. A model for niemann-pick type C disease in the nematode *Caenorhabditis elegans*. Curr. Biol. 10: 527-530.
4. Cruz, J.C., et al. 2000. Fate of endogenously synthesized cholesterol in Niemann-Pick type C1 cells. J. Biol. Chem. 275: 41309-41316.
5. Ioannou, Y.A. 2000. The structure and function of the Niemann-Pick C1 protein. Mol. Genet. Metab. 71: 175-181.
6. Watabe, K., et al. 2001. Establishment and characterization of immortalized Schwann cells from murine model of Niemann-Pick disease type C (spm/spm). J. Peripher. Nerv. Syst. 6: 85-94.
7. Sun, X., et al. 2001. Niemann-Pick C variant detection by altered sphingolipid trafficking and correlation with mutations within a specific domain of NPC1. Am. J. Hum. Genet. 68: 1361-1372.
8. Zervas, M., et al. 2001. Neurons in Niemann-Pick disease type C accumulate gangliosides as well as unesterified cholesterol and undergo dendritic and axonal alterations. J. Neuropathol. Exp. Neurol. 60: 49-64.

CHROMOSOMAL LOCATION

Genetic locus: NPC1 (human) mapping to 18q11.2; Npc1 (mouse) mapping to 18 A1.

SOURCE

NPC1 (G-1) is a mouse monoclonal antibody raised against amino acids 516-630 of NPC1 of human origin.

PRODUCT

Each vial contains 200 µg IgA kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

NPC1 (G-1) is recommended for detection of NPC1 of mouse, rat and human origin by Western Blotting (starting dilution 1:100, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for NPC1 siRNA (h): sc-41588, NPC1 siRNA (m): sc-41589, NPC1 shRNA Plasmid (h): sc-41588-SH, NPC1 shRNA Plasmid (m): sc-41589-SH, NPC1 shRNA (h) Lentiviral Particles: sc-41588-V and NPC1 shRNA (m) Lentiviral Particles: sc-41589-V.

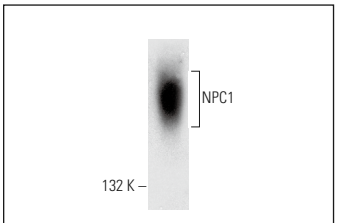
Molecular Weight of glycosylated NPC1: 170/190 kDa.

Positive Controls: ES-2 cell lysate: sc-24674.

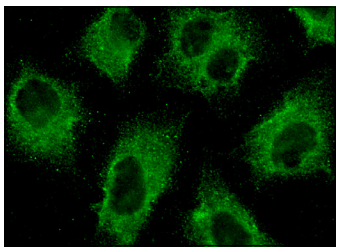
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein L-Agarose: sc-2336 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

DATA



NPC1 (G-1): sc-271335. Western blot analysis of NPC1 expression in ES-2 whole cell lysate.



NPC1 (G-1): sc-271335. Immunofluorescence staining of methanol-fixed HeLa cells showing cytoplasmic localization.

SELECT PRODUCT CITATIONS

1. Lin, J.X., et al. 2023. Rab7a-mTORC1 signaling-mediated cholesterol trafficking from the lysosome to mitochondria ameliorates hepatic lipotoxicity induced by aflatoxin B1 exposure. Chemosphere 320: 138071.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

For research use only, not for use in diagnostic procedures.