

NPC1 (E-9): sc-271334

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

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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.
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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.

SOURCE

NPC1 (E-9) is a mouse monoclonal antibody raised against amino acids 516-630 of NPC1 of human origin.

PRODUCT

Each vial contains 200 µg IgG_{2a} kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

NPC1 (E-9) is recommended for detection of NPC1 of 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 shRNA Plasmid (h): sc-41588-SH and NPC1 shRNA (h) Lentiviral Particles: sc-41588-V.

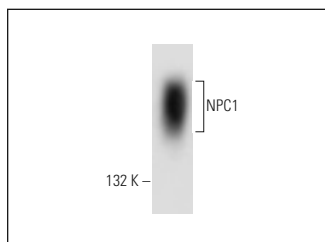
Molecular Weight of glycosylated NPC1: 170/190 kDa.

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 A/G PLUS-Agarose: sc-2003 (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 (E-9): sc-271334. Western blot analysis of NPC1 expression in ES-2 whole cell lysate.

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.

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.