

# NIPA1 (E-4): sc-398041

## BACKGROUND

NIPA1 (non imprinted in Prader-Willi/Angelman syndrome 1), also known as SPG6 or FSP3, is a 329 amino acid multi-pass membrane protein that exists as multiple alternatively spliced isoforms and is expressed at high levels in neuronal tissue. NIPA1 is thought to play a role in nervous system development and, when defective, is involved in the pathogenesis of spastic paraplegia autosomal dominant type 6 (SPG6), a degenerative spinal cord disease characterized by the progressive weakening of the lower limbs. The gene encoding NIPA1 maps to human chromosome 15, which houses over 700 genes and comprises nearly 3% of the human genome. Angelman syndrome, Prader-Willi syndrome, Tay-Sachs disease and Marfan syndrome are all associated with defects in chromosome 15-localized genes.

## REFERENCES

1. Fink, J.K., et al. 1995. Autosomal dominant familial spastic paraplegia: tight linkage to chromosome 15q. *Am. J. Hum. Genet.* 56: 188-192.
2. Fink, J.K., et al. 1995. Autosomal dominant, familial spastic paraplegia, type I: clinical and genetic analysis of a large North American family. *Neurology* 45: 325-331.
3. Chai, J.H., et al. 2003. Identification of four highly conserved genes between breakpoint hotspots BP1 and BP2 of the Prader-Willi/Angelman syndromes deletion region that have undergone evolutionary transposition mediated by flanking duplicons. *Am. J. Hum. Genet.* 73: 898-925.
4. Rainier, S., et al. 2003. NIPA1 gene mutations cause autosomal dominant hereditary spastic paraplegia (SPG6). *Am. J. Hum. Genet.* 73: 967-971.
5. Chen, S., et al. 2005. Distinct novel mutations affecting the same base in the NIPA1 gene cause autosomal dominant hereditary spastic paraplegia in two Chinese families. *Hum. Mutat.* 25: 135-141.
6. Reed, J.A., et al. 2005. A novel NIPA1 mutation associated with a pure form of autosomal dominant hereditary spastic paraplegia. *Neurogenetics* 6: 79-84.

## CHROMOSOMAL LOCATION

Genetic locus: NIPA1 (human) mapping to 15q11.2; Nipa1 (mouse) mapping to 7 B5.

## SOURCE

NIPA1 (E-4) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 210-229 within a cytoplasmic domain of NIPA1 of human origin.

## PRODUCT

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

Blocking peptide available for competition studies, sc-398041 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% stabilizer protein).

## APPLICATIONS

NIPA1 (E-4) is recommended for detection of NIPA1 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 NIPA1 siRNA (h): sc-89918, NIPA1 siRNA (m): sc-106304, NIPA1 siRNA (r): sc-270012, NIPA1 shRNA Plasmid (h): sc-89918-SH, NIPA1 shRNA Plasmid (m): sc-106304-SH, NIPA1 shRNA Plasmid (r): sc-270012-SH, NIPA1 shRNA (h) Lentiviral Particles: sc-89918-V, NIPA1 shRNA (m) Lentiviral Particles: sc-106304-V and NIPA1 shRNA (r) Lentiviral Particles: sc-270012-V.

Molecular Weight (predicted) of NIPA1 isoforms: 35/27 kDa.

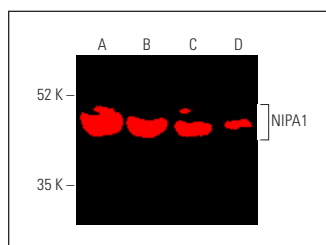
Molecular Weight (observed) of NIPA1: 43 kDa.

Positive Controls: rat brain extract: sc-2392, mouse brain extract: sc-2253 or rat cerebellum extract: sc-2398.

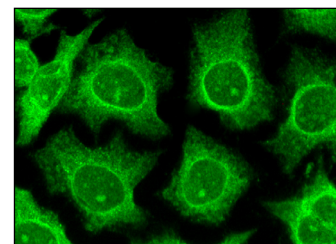
## RECOMMENDED SUPPORT REAGENTS

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



NIPA1 (E-4): sc-398041. Near-infrared western blot analysis of NIPA1 expression in rat cerebellum (A), mouse brain (B), rat brain (C) and human brain (D) tissue extracts. Blocked with UltraCruz® Blocking Reagent: sc-516214. Detection reagent used: m-IgGκ BP-CFL 790: sc-516181.



NIPA1 (E-4): sc-398041. Immunofluorescence staining of methanol-fixed HeLa cells showing cytoplasmic and membrane localization.

## 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.