SANTA CRUZ BIOTECHNOLOGY, INC.

Malin (N-19): sc-54234

BACKGROUND

Progressive myoclonic epilepsy type 2 (EPM2), also called Lafora disease, is an autosomal recessive disease characterized by grand mal seizures and/or myoclonus at about 15 years of age. Rapid and severe mental deterioration follows, often with psychotic features. Survival is less than 10 years after onset. Starch-like, endoplasmic reticulum-associated polyglucosans, called Lafora bodies, can be observed in brain, muscle, liver and heart. One cause of Lafora disease is due to mutations in NHLRC1, the gene encoding Malin. 49 different mutations in NHLRC1 have been shown to cause EPM2. Malin, also called NHL repeat-containing protein 1, is a single subunit E3 ubiquitin ligase, containing six NHL repeats and one RING-type zinc finger. Malin's RING domain is responsible for its ability to mediate ubiquitination. Malin interacts with and polyubiquitinates Laforin, a protein also implicated in EPM2. Malin localizes to the endoplasmic reticulum and, to a lesser extent, in the nucleus. Malin is expressed in brain, cerebellum, spinal cord, medulla, heart, liver, skeletal muscle and pancreas.

REFERENCES

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2. Chan, E.M., et al. 2004. Progressive myoclonus epilepsy with polyglucosans (Lafora disease): evidence for a third locus. Neurology 63: 565-567.
3. Ianzano, L., et al. 2005. Lafora progressive Myoclonus Epilepsy mutation database-EPM2A and NHLRC1 (EPM2B) genes. Hum. Mutat. 26: 397-397.
4. Lohi, H., et al. 2005. Novel glycogen synthase kinase 3 and ubiquitination pathways in progressive myoclonus epilepsy. Hum. Mol. Genet. 14: 2727-2736.
5. Gentry, M.S., et al. 2005. Insights into Lafora disease: malin is an E3 ubiquitin ligase that ubiquitinates and promotes the degradation of laforin. Proc. Natl. Acad. Sci. USA 102: 8501-8506.
6. Ganesh, S., et al. 2006. Recent advances in the molecular basis of Lafora's progressive myoclonus epilepsy. J. Hum. Genet. 51: 1-8.
7. Singh, S., et al. 2006. Novel NHLRC1 mutations and genotype-phenotype correlations in patients with Lafora's progressive myoclonic epilepsy. J. Med. Genet. 43: e48.
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CHROMOSOMAL LOCATION

Genetic locus: NHLRC1 (human) mapping to 6p22.3.

SOURCE

Malin (N-19) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the N-terminus of Malin of human origin.

STORAGE

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

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-54234 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

Malin (N-19) is recommended for detection of Malin of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), 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 Malin siRNA (h): sc-106193, Malin shRNA Plasmid (h): sc-106193-SH and Malin shRNA (h) Lentiviral Particles: sc-106193-V.

Molecular Weight of Malin: 42 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

RESEARCH USE

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

PROTOCOLS

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