SANTA CRUZ BIOTECHNOLOGY, INC.

Malin (3G6): sc-293401

SANTA CRUZ BIOTECHNOLOGY

The Power to Question

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 ten 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. Forty-nine 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 1 RING-type zinc finger. The RING domain of Malin 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.
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.
8. Mittal, S., et al. 2007. Lafora disease proteins malin and laforin are recruited to aggresomes in response to proteasomal impairment. Hum. Mol. Genet. 16: 753-762.

CHROMOSOMAL LOCATION

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

SOURCE

Malin (3G6) is a mouse monoclonal antibody raised against amino acids 137-246 of Malin of human origin.

PRODUCT

Each vial contains 100 µg IgG_{2a} kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

Malin (3G6) is recommended for detection of Malin of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)] and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

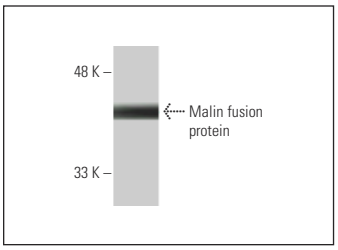
Suitable for use as control antibody for Malin siRNA (h): sc-106193, Malin shRNA Plasmid (h): sc-106193-SH and Malin shRNA (h) Lentiviral Particles: sc-106193-V.

Molecular Weight of Malin: 42 kDa.

RECOMMENDED 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).

DATA



Malin (3G6): sc-293401. Western blot analysis of human recombinant Malin fusion protein.

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.