SANTA CRUZ BIOTECHNOLOGY, INC.

myotubularin (E-16): sc-14783

SANTA CRUZ BIOTECHNOLOGY

The Power to Question

BACKGROUND

X-linked recessive myotubular myopathy is a congenital muscular disease characterized by severe hypotonia and generalized muscle weakness that, in most cases, leads to early postnatal death. The gene responsible for myotubular myopathy MTM1 encodes a dual specificity phosphatase, named myotubularin, which is highly conserved through evolution. The gene for MTM1 is localized to a 300 kb critical region on human Xq128 between IDS and GRBRA3. Human MTM1, a 603 amino-acid protein, is mutated in myotubular myopathy. The largely related protein hMTMR2 is found mutated in a recessive form of Charcot-Marie-Tooth neuropathy. Myotubularin is primarily a lipid phosphatase that acts on phosphatidylinositol 3-monophosphate and is involved in the regulation of the phosphatidylinositol 3-kinase (PI3-kinase) pathway and membrane trafficking. Wild-type myotubularin can directly dephosphorylate PI3P and PI4P *in vitro*. Thus, it decreases PI3P levels by down-regulating PI3K activity and by facilitating the degradation of PI3P.

REFERENCES

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2. de Gouyon, B., et al. 1996. Comparative mapping on the mouse X chromosome defines a myotubular myopathy equivalent region. Mamm. Genome 7: 575-579.
3. Laporte, J., et al. 1997. Mutations in the MTM1 gene implicated in X-llinked myotubular myopathy. Hum. Mol. Genet. 6: 1505-1511.
4. Buj-Bello, A., et al. 1999. Identification of novel mutations in the MTM1 gene causing severe and mild forms of X-linked mytotubular myopathy. Hum. Mutat. 14: 320-325.
5. Hane, B.G., et al. 1999. Germline mosaicism in X-linked myotubular myopathy. Clin. Genet. 56: 77-81.
6. Blondeau, F., et al. 2000. Myotubularin, a phosphatase deficient in myotubular myopathy, acts on phosphatidylinositol 3-kinase and phosphatidylinositol 3-phosphate pathway. Hum. Mol. Genet. 9: 2223-2229.
7. Laporte, J., et al. 2001. The myotubularin family: from genetic disease to phosphoinositide metabolism. Trends Genet. 17: 221-228.

CHROMOSOMAL LOCATION

Genetic locus: MTM1 (human) mapping to Xq28; Mtm1 (mouse) mapping to X A7.2.

SOURCE

myotubularin (E-16) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the C-terminus of myotubularin 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-14783 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

myotubularin (E-16) is recommended for detection of myotubularin of mouse, rat and 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).

myotubularin (E-16) is also recommended for detection of myotubularin in additional species, including equine, canine, bovine and porcine.

Suitable for use as control antibody for myotubularin siRNA (h): sc-44356, myotubularin siRNA (m): sc-44357, myotubularin shRNA Plasmid (h): sc-44356-SH, myotubularin shRNA Plasmid (m): sc-44357-SH, myotubularin shRNA (h) Lentiviral Particles: sc-44356-V and myotubularin shRNA (m) Lentiviral Particles: sc-44357-V.

Molecular Weight of myotubularin: 66 kDa.

Positive Controls: Hep G2 cell lysate: sc-2227.

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.