SANTA CRUZ BIOTECHNOLOGY, INC.

myotubularin (Y-12): sc-100399

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 (PI 3-kinase) pathway and membrane trafficking. Wildtype myotubularin can directly dephosphorylate PI 3-P and PI 4-P *in vitro*. Thus, it decreases PI 3-P levels by down-regulating PI 3-K activity and by facilitating the degradation of PI 3-P.

REFERENCES

1. Laporte, J., et al. 1996. A gene mutated in X-linked myotubular myopathy defines a new putative tyrosine phosphatase family conserved in yeast. Nat. Genet. 13: 175-182.
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-linked 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.

STORAGE

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

PROTOCOLS

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

SOURCE

myotubularin (Y-12) is a mouse monoclonal antibody raised against recombinant myotubularin of human origin.

PRODUCT

Each vial contains 50 µg IgG_{2a} in 500 µl PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

myotubularin (Y-12) is recommended for detection of myotubularin of human origin by immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500), immunohistochemistry (including paraffin-embedded sections) (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 myotubularin siRNA (h): sc-44356, myotubularin shRNA Plasmid (h): sc-44356-SH and myotubularin shRNA (h) Lentiviral Particles: sc-44356-V.

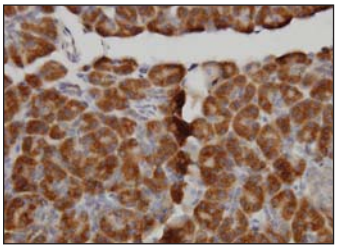
Molecular Weight of myotubularin: 66 kDa.

Positive Controls: human pancreas tissue.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Immunofluorescence: use goat anti-mouse IgG-FITC: sc-2010 (dilution range: 1:100-1:400) or goat anti-mouse IgG-TR: sc-2781 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941. 2) Immunohistochemistry: use ImmunoCruz™: sc-2050 or ABC: sc-2017 mouse IgG Staining Systems.

DATA



myotubularin (Y-12): sc-100399. Immunoperoxidase staining of formalin-fixed, paraffin-embedded human pancreas tissue showing cytoplasmic localization.

RESEARCH USE

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