SANTA CRUZ BIOTECHNOLOGY, INC.

myotubularin (F-1): sc-377309

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 critical region on human Xq28 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

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.

CHROMOSOMAL LOCATION

Genetic locus: MTM1 (human) mapping to Xq28.

SOURCE

myotubularin (F-1) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 21-57 near the N-terminus of myotubularin of human origin.

PRODUCT

Each vial contains 200 µg IgG_{2b} kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

myotubularin (F-1) is available conjugated to agarose (sc-377309 AC), 500 µg/0.25 ml agarose in 1 ml, for IP; to HRP (sc-377309 HRP), 200 µg/ml, for WB, IHC(P) and ELISA; to either phycoerythrin (sc-377309 PE), fluorescein (sc-377309 FITC), Alexa Fluor® 488 (sc-377309 AF488), Alexa Fluor® 546 (sc-377309 AF546), Alexa Fluor® 594 (sc-377309 AF594) or Alexa Fluor® 647 (sc-377309 AF647), 200 µg/ml, for WB (RGB), IF, IHC(P) and FCM; and to either Alexa Fluor® 680 (sc-377309 AF680) or Alexa Fluor® 790 (sc-377309 AF790), 200 µg/ml, for Near-Infrared (NIR) WB, IF and FCM.

Blocking peptide available for competition studies, sc-377309 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% stabilizer protein).

RESEARCH USE

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

APPLICATIONS

myotubularin (F-1) is recommended for detection of myotubularin of 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), 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.

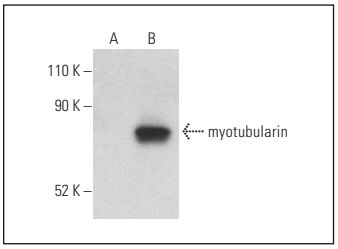
Molecular Weight of myotubularin: 66 kDa.

Positive Controls: Hep G2 cell lysate: sc-2227 or myotubularin (h): 293 Lysate: sc-158751.

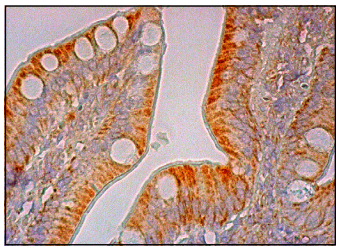
RECOMMENDED SUPPORT REAGENTS

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). 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. 4) Immunohistochemistry: use m-IgGκ BP-HRP: sc-516102 with DAB, 50X: sc-24982 and Immunohistomount: sc-45086, or Organo/Limonene Mount: sc-45087.

DATA



myotubularin (F-1): sc-377309. Western blot analysis of myotubularin expression in non-transfected: sc-110760 (**A**) and human myotubularin transfected: sc-158751 (**B**) 293 whole cell lysates. Detection reagent used: m-IgG Fc BP-HRP: sc-525409.



myotubularin (F-1): sc-377309. Immunoperoxidase staining of formalin fixed, paraffin-embedded human duodenum tissue showing cytoplasmic staining of glandular cells.

SELECT PRODUCT CITATIONS

1. Olby, N.J., et al. 2020. A mutation in MTM1 causes X-Linked myotubular myopathy in Boykin spaniels. Neuromuscul. Disord. 30: 353-359.

STORAGE

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