SANTA CRUZ BIOTECHNOLOGY, INC.

MTMR1 (1B9): sc-134395

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. 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. Wild-type myotubularin can directly dephosphorylate PI3P and PI4P *in vitro.* Thus, it decreases PI3P levels by downregulating PI 3-kinase activity and by facilitating the degradation of PI3P. The MTMR1 gene is adjacent to MTM1 on chromosome X, and its protein shares 59% sequence identity with Myotubularin. MTMR1 also plays a role in muscle formation and is deleted in patients with myotubular myopathy.

REFERENCES

1. Laporte, J., et al. 1996. A gene mutated in X-linked myotubular myopathy defines a new putative tyrosine phosphatase family conserved inyeast. Nat. Genet. 13: 175-182.
2. Laporte, J., et al. 1997. Mutations in the MTM1 gene implicated in X-llinked myotubular myopathy. Hum. Mol. Genet. 6: 1505-1511.
3. 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.
4. 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.
5. Buj-Bello, A., et al. 2002. Muscle-specific alternative splicing of myotubularin-related 1 gene is impaired in DM1 muscle cells. Hum. Mol. Genet. 11: 2297-2307.
6. Zanoteli, E., et al. 2005. Deletion of both MTM1 and MTMR1 genes in a boy with myotubular myopathy. Am. J. Med. Genet. A 134: 338-340.

CHROMOSOMAL LOCATION

Genetic locus: MTMR1 (human) mapping to Xq28; Mtmr1 (mouse) mapping to X A7.2.

SOURCE

MTMR1 (1B9) is a mouse monoclonal antibody raised against recombinant MTMR1 protein 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.

STORAGE

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

APPLICATIONS

MTMR1 (1B9) is recommended for detection of MTMR1 of mouse, rat and 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 MTMR1 siRNA (h): sc-61084, MTMR1 siRNA (m): sc-61085, MTMR1 shRNA Plasmid (h): sc-61084-SH, MTMR1 shRNA Plasmid (m): sc-61085-SH, MTMR1 shRNA (h) Lentiviral Particles: sc-61084-V and MTMR1 shRNA (m) Lentiviral Particles: sc-61085-V.

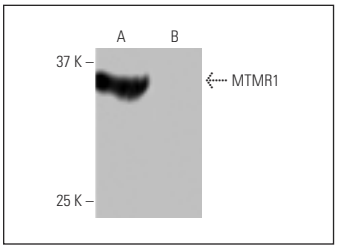
Molecular Weight of MTMR1: 75 kDa.

Positive Controls: human MTMR1 transfected 293T whole cell lysate.

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



MTMR1 (1B9): sc-134395. Western blot analysis of MTMR1 expression in human MTMR1 transfected (**A**) and non-transfected (**B**) 293T whole cell lysates.

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.