

myotubularin (N-20): sc-14781

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 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 (PI 3-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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- de Gouyon, B., et al. 1996. Comparative mapping on the mouse X chromosome defines a myotubular myopathy equivalent region. *Mamm. Genome* 7: 575-579.
- Laporte, J., et al. 1997. Mutations in the MTM1 gene implicated in X-linked myotubular myopathy. *Hum. Mol. Genet.* 6: 1505-1511.
- Buj-Bello, A., et al. 1999. Identification of novel mutations in the MTM1 gene causing severe and mild forms of X-linked myotubular myopathy. *Hum. Mutat.* 14: 320-325.
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- 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.
- 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 (N-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-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-14781 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

myotubularin (N-20) is recommended for detection of myotubularin of human and, to a lesser extent, mouse and rat 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)], 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).

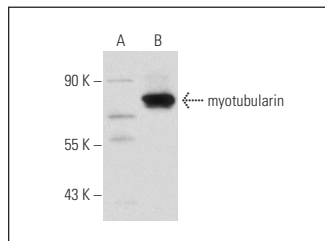
myotubularin (N-20) is also recommended for detection of myotubularin in additional species, including equine, canine, bovine and porcine.

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.

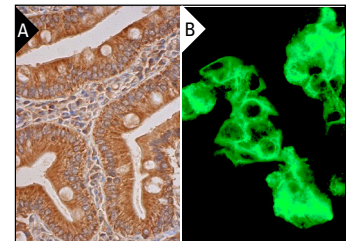
Molecular Weight of myotubularin: 66 kDa.

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

DATA



myotubularin (N-20): sc-14781. Western blot analysis of myotubularin expression in non-transfected: sc-110760 (A) and human myotubularin transfected: sc-158751 (B) 293 whole cell lysates.



myotubularin (N-20): sc-14781. Immunoperoxidase staining of formalin fixed, paraffin-embedded human duodenum tissue showing cytoplasmic staining of glandular cells (A). Immunofluorescence staining of methanol-fixed Hep G2 cells showing membrane and cytoplasmic localization (B).

RESEARCH USE

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


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Try **myotubularin (F-1): sc-377309**, our highly recommended monoclonal alternative to myotubularin (N-20).