SANTA CRUZ BIOTECHNOLOGY, INC.

myotubularin (h): 293T Lysate: sc-158751

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.
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.

PRODUCT

myotubularin (h): 293T Lysate represents a lysate of human myotubularin transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

APPLICATIONS

myotubularin (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive myotubularin antibodies. Recommended use: 10-20 µl per lane.

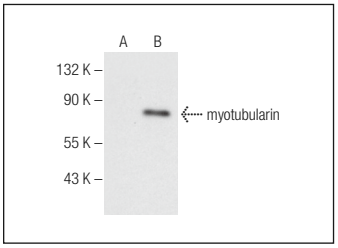
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

myotubularin (F-1): sc-377309 is recommended as a positive control antibody for Western Blot analysis of enhanced human myotubularin expression in myotubularin transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

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.

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.

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.