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SZABO-SCANDIC HandelsgmbH

Quellenstraße 110, A-1100 Wien

T. +43(0)1 489 3961-0

F. +43(0)1 489 3961-7

mail@szabo-scandic.com

www.szabo-scandic.com

[linkedin.com/company/szaboscandic](https://www.linkedin.com/company/szaboscandic) 

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 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. 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

- 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.
- 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.
- Hane, B.G., et al. 1999. Germline mosaicism in X-linked myotubular myopathy. *Clin. Genet.* 56: 77-81.
- 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.

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.

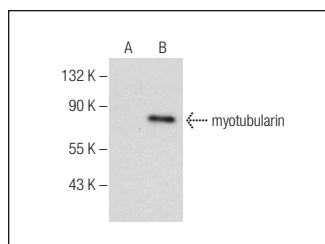
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

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.