SANTA CRUZ BIOTECHNOLOGY, INC.

# MTMR1 siRNA (h): sc-61084

## 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 (PI3-kinase) pathway and membrane trafficking. Wildtype 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. 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 in yeast. Nat. Genet. 13: 175-182.
2. Laporte, J., et al. 1997. Mutations in the MTM1 gene implicated in X-linked 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.

## PRODUCT

MTMR1 siRNA (h) is a pool of 3 target-specific 19-25 nt siRNAs designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10 µM solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see MTMR1 shRNA Plasmid (h): sc-61084-SH and MTMR1 shRNA (h) Lentiviral Particles: sc-61084-V as alternate gene silencing products.

For independent verification of MTMR1 (h) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-61084A, sc-61084B and sc-61084C.

## RESEARCH USE

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

## STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20° C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20° C, avoid contact with RNAses and repeated freeze thaw cycles.

Resuspend lyophilized siRNA duplex in 330 µl of the RNAse-free water provided. Resuspension of the siRNA duplex in 330 µl of RNAse-free water makes a 10 µM solution in a 10 µM Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

## APPLICATIONS

MTMR1 siRNA (h) is recommended for the inhibition of MTMR1 expression in human cells.

## SUPPORT REAGENTS

For optimal siRNA transfection efficiency, Santa Cruz Biotechnology's siRNA Transfection Reagent: sc-29528 (0.3 ml), siRNA Transfection Medium: sc-36868 (20 ml) and siRNA Dilution Buffer: sc-29527 (1.5 ml) are recommended. Control siRNAs or Fluorescein Conjugated Control siRNAs are available as 10 µM in 66 µl. Each contain a scrambled sequence that will not lead to the specific degradation of any known cellular mRNA. Fluorescein Conjugated Control siRNAs include: sc-36869, sc-44239, sc-44240 and sc-44241. Control siRNAs include: sc-37007, sc-44230, sc-44231, sc-44232, sc-44233, sc-44234, sc-44235, sc-44236, sc-44237 and sc-44238.

## GENE EXPRESSION MONITORING

MTMR1 (1B9): sc-134395 is recommended as a control antibody for monitoring of MTMR1 gene expression knockdown by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000) or immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500).

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) 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.

## RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor MTMR1 gene expression knockdown using RT-PCR Primer: MTMR1 (h)-PR: sc-61084-PR (20 µl). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

## PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.