



mucolipin 1 siRNA (h): sc-44519

BACKGROUND

The gene encoding human mucolipin 1 maps to chromosome 19p13.2. Mutations in this gene cause a rare autosomal recessive lysosomal storage disease known as mucopolipidosis type IV (MLIV). Clinical characteristics of MLIV include psychomotor retardation, retinal degeneration, corneal opacities and strabismus. Mucolipin 1 localizes to the plasma membrane and contains six transmembrane domains. The carboxy terminus of mucolipin 1 shares sequence homology with Polycystin-2 and the transient receptor potential cation channel family. The concentration of intracellular Ca^{2+} regulates the permeability of mucolipin 1 to Ca^{2+} , Na^{+} and K^{+} . The influence of Ca^{2+} on mucolipin 1 represents a possible role for mucolipin 1 in lysosomal exocytosis and the trafficking of late endosomes and lysosomes.

REFERENCES

- Merin, S., et al. 1975. Mucopolipidosis IV: ocular, systemic and ultrastructural findings. *Invest. Ophthalmol.* 14: 437-448.
- Bargal, R., et al. 2000. Identification of the gene causing mucopolipidosis type IV. *Nat. Genet.* 26: 118-123.
- Sun, M., et al. 2000. Mucopolipidosis type IV is caused by mutations in a gene encoding a novel transient receptor potential channel. *Hum. Mol. Genet.* 9: 2471-2478.
- Bassi, M.T., et al. 2000. Cloning of the gene encoding a novel integral membrane protein, mucolipidin and identification of the two major founder mutations causing mucopolipidosis type IV. *Am. J. Hum. Genet.* 67: 1110-1120.

CHROMOSOMAL LOCATION

Genetic locus: MCOLN1 (human) mapping to 19p13.2.

PRODUCT

mucolipin 1 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 mucolipin 1 shRNA Plasmid (h): sc-44519-SH and mucolipin 1 shRNA (h) Lentiviral Particles: sc-44519-V as alternate gene silencing products.

For independent verification of mucolipin 1 (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-44519A, sc-44519B and sc-44519C.

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

mucolipin 1 siRNA (h) is recommended for the inhibition of mucolipin 1 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

mucolipin 1 (F-10): sc-398868 is recommended as a control antibody for monitoring of mucolipin 1 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-IgG κ BP-HRP: sc-516102 or m-IgG κ 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-IgG κ BP-FITC: sc-516140 or m-IgG κ 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 mucolipin 1 gene expression knockdown using RT-PCR Primer: mucolipin 1 (h)-PR: sc-44519-PR (20 μl , 467 bp). Annealing temperature for the primers should be $55-60^{\circ}\text{C}$ and the extension temperature should be $68-72^{\circ}\text{C}$.

SELECT PRODUCT CITATIONS

- Xu, Y., et al. 2021. TFEB regulates lysosomal exocytosis of tau and its loss of function exacerbates Tau pathology and spreading. *Mol. Psychiatry* 26: 5925-5939.
- Nabar, N.R., et al. 2022. LRRK2 is required for CD38-mediated NAADP- Ca^{2+} signaling and the downstream activation of TFEB (transcription factor EB) in immune cells. *Autophagy* 18: 204-222.
- Goretzko, J., et al. 2023. P-selectin-dependent leukocyte adhesion is governed by endolysosomal two-pore channel 2. *Cell Rep.* 42: 113501.

RESEARCH USE

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