



# SZABO SCANDIC

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Diagnostik & molekulare Diagnostik



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# mucolipin 1 (h): 293T Lysate: sc-171244

## 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

- Bargal, R., Avidan, N., Ben-Asher, E., Olender, Z., Zeigler, M., Frumkin, A., Raas-Rothschild, A., Glusman, G., Lancet, D. and Bach, G. 2000. Identification of the gene causing mucopolipidosis type IV. *Nat. Genet.* 26: 118-123.
- Merin, S., Livni, N., Berman, E.R. and Yatziv, S. 1975. Mucopolipidosis IV: ocular, systemic, and ultrastructural findings. *Invest. Ophthalmol.* 14: 437-448.
- Sun, M., Goldin, E., Stahl, S., Falardeau, J.L., Kennedy, J.C., Acierno, J.S., Jr., Bove, C., Kaneski, C.R., Nagle, J., Bromley, M.C., Colman, M., Schiffmann, R. and Slaugenhaupt, S.A. 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., Manzoni, M., Monti, E., Pizzo, M.T., Ballabio, A. and Borsani, G. 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.
- LaPlante, J.M., Falardeau, J., Sun, M., Kanazirska, M., Brown, E.M., Slaugenhaupt, S.A. and Vassilev, P.M. 2002. Identification and characterization of the single channel function of human mucolipin-1 implicated in mucopolipidosis type IV, a disorder affecting the lysosomal pathway. *FEBS Lett.* 532: 183-187.

## CHROMOSOMAL LOCATION

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

## PRODUCT

mucolipin 1 (h): 293T Lysate represents a lysate of human mucolipin 1 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.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) for detailed protocols and support products.

## APPLICATIONS

mucolipin 1 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive mucolipin 1 antibodies. Recommended use: 10-20 µl per lane.

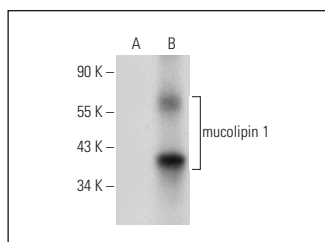
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

mucolipin 1 (F-10): sc-398868 is recommended as a positive control antibody for Western Blot analysis of enhanced human mucolipin 1 expression in mucolipin 1 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



mucolipin 1 (F-10): sc-398868. Western blot analysis of mucolipin 1 expression in non-transfected: sc-117752 (A) and human mucolipin 1 transfected: sc-171244 (B) 293T whole cell lysates.

## RESEARCH USE

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