SANTA CRUZ BIOTECHNOLOGY, INC.

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 mucolipidosis 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 lysosmes.

REFERENCES

1. Merin, S., Livni, N., Berman, E.R. and Yatziv, S. 1975. Mucolipidosis IV: ocular, systemic, and ultrastructural findings. Invest. Ophthalmol. 14: 437-448.
2. 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 mucolipidosis type IV. Nat. Genet. 26: 118-123.
3. 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. Mucolipidosis type IV is caused by mutations in a gene encoding a novel transient receptor potential channel. Hum. Mol. Genet. 9: 2471-2478.
4. 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 mucolipidosis type IV. Am. J. Hum. Genet. 67: 1110-1120.
5. 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 mucolipidosis 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.

RESEARCH USE

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

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.

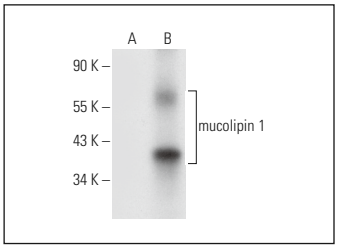
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

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.

PROTOCOLS

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