

# mucolipin 1 (M-13): sc-26269

## 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.
- Bassi, M.T., et al. 2000. Cloning of the gene encoding a novel integral membrane protein, mucolipin and identification of the two major founder mutations causing mucopolipidosis type IV. *Am. J. Hum. Genet.* 67:

## CHROMOSOMAL LOCATION

Genetic locus: MCOLN1 (human) mapping to 19p13.2; Mcoln1 (mouse) mapping to 8 A1.1.

## SOURCE

mucolipin 1 (M-13) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the C-terminus of mucolipin 1 of mouse origin.

## PRODUCT

Each vial contains 200  $\mu$ g IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-26269 P, (100  $\mu$ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

mucolipin 1 (M-13) is recommended for detection of mucolipin 1 of mouse, rat and, to a lesser extent, human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500), immunohistochemistry (including paraffin-embedded sections) (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for mucolipin 1 siRNA (h): sc-44519, mucolipin 1 siRNA (m): sc-44520, mucolipin 1 shRNA Plasmid (h): sc-44519-SH, mucolipin 1 shRNA Plasmid (m): sc-44520-SH, mucolipin 1 shRNA (h) Lentiviral Particles: sc-44519-V and mucolipin 1 shRNA (m) Lentiviral Particles: sc-44520-V.

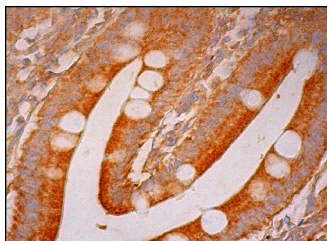
Molecular Weight of mucolipin: 65 kDa.

Positive Controls: mouse PBL whole cell lysate.

## RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941. 3) Immunohistochemistry: use ImmunoCruz™: sc-2053 or ABC: sc-2023 goat IgG Staining Systems.

## DATA



mucolipin 1 (M-13): sc-26269. Immunoperoxidase staining of formalin fixed, paraffin-embedded human small intestine tissue showing cytoplasmic staining of glandular cells.

## SELECT PRODUCT CITATIONS

- Zhang, F. and Li, P.L. 2007. Reconstitution and characterization of a nicotinic acid adenine dinucleotide phosphate (NAADP)-sensitive  $Ca^{2+}$  release channel from liver lysosomes of rats. *J. Biol. Chem.* 282: 25259-25269.
- Zhang, F., et al. 2008. TRP-ML1 functions as a lysosomal NAADP-sensitive  $Ca^{2+}$  release channel in coronary arterial myocytes. *J. Cell. Mol. Med.* 13: 3174-3185.

## STORAGE

Store at 4° C, \*\*DO NOT FREEZE\*\*. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

## RESEARCH USE

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

## PROTOCOLS

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

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Try **mucolipin 1 (F-10): sc-398868**, our highly recommended monoclonal alternative to mucolipin 1 (M-13).