

mucolipin 1 (H-75): sc-48745

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.

CHROMOSOMAL LOCATION

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

SOURCE

mucolipin 1 (H-75) is a rabbit polyclonal antibody raised against amino acids 106-180 mapping within an internal region of mucolipin 1 of human origin.

PRODUCT

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

APPLICATIONS

mucolipin 1 (H-75) is recommended for detection of mucolipin 1 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 μ g per 100-500 μ g of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

mucolipin 1 (H-75) is also recommended for detection of mucolipin 1 in additional species, including equine, canine, bovine and porcine.

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.

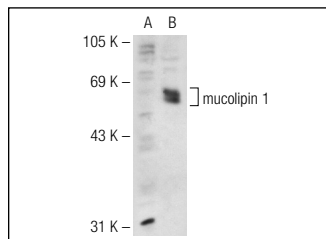
Molecular Weight of mucolipin: 65 kDa.

Positive Controls: mucolipin 1 (h2): 293T Lysate: sc-172042 or Jurkat whole cell lysate: sc-2204.

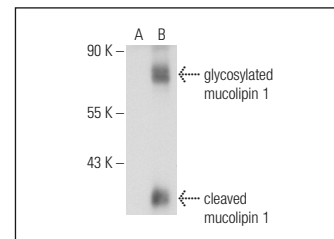
RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

DATA



mucolipin 1 (H-75): sc-48745. 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.



mucolipin 1 (H-75): sc-48745. Western blot analysis of mucolipin 1 expression in non-transfected: sc-117752 (A) and human mucolipin 1 transfected: sc-172042 (B) 293T whole cell lysates.

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 or our catalog for detailed protocols and support products.

MONOS
Satisfaction
Guaranteed

Try **mucolipin 1 (F-10): sc-398868**, our highly recommended monoclonal alternative to mucolipin 1 (H-75).