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SANTA CRUZ BIOTECHNOLOGY, INC.

mucolipin 1 (h2): 293T Lysate: sc-172042

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

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CHROMOSOMAL LOCATION

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

PRODUCT

mucolipin 1 (h2): 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 for detailed protocols and support products.

APPLICATIONS

mucolipin 1 (h2): 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.

RESEARCH USE

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