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Product Information

Monoclonal Anti-Mucolipin-1, Clone MLN128
produced in mouse, purified immunoglobulin

Product Number **M8072**

Product Description

Monoclonal Anti-Mucolipin-1 (mouse IgG1 isotype) is derived from the hybridoma MLN128 produced by the fusion of mouse myeloma cells and splenocytes from BALB/c mice immunized with a recombinant fusion protein corresponding to a fragment of human Mucolipin-1 (GeneID 57192). The isotype is determined using a double diffusion immunoassay using Mouse Monoclonal Antibody Isotyping Reagents, Product Number ISO2.

Monoclonal Anti-Mucolipin-1 recognizes human mucolipin-1. The antibody may be used in various immunochemical techniques including ELISA, immunoblotting (~110 kDa), and immunocytochemistry. Endogenous Mucolipin-1 (~65 kDa) may undergo several processes, such as cleavage, e.g., 35-40 kDa N-terminal cleavage product. Therefore, depending on the cell source and treatment, additional bands may be observed.[1]

Mucolipidosis type IV (MLIV) is an autosomal recessive, neurodegenerative disorder caused by mutations in the *MCOLN1* gene that encodes mucolipin-1 (also termed TRP-ML1, MLN1, ML1, mucolipidin).[2,3] MLIV is associated with severe psychomotor retardation and ophthalmologic defects. MLIV is a lysosomal storage disorder associated with lysosomal accumulation of sphingolipids, phospholipids, and mucopolysaccharides. Unlike other mucolipidoses, lysosomal hydrolase activity is not impaired in MLIV. Rather, MLIV pathophysiology has been linked to deficiency in membrane trafficking, and organelle dynamics in the late endocytic pathway. Specifically, MLIV cells have been shown to accumulate autophagosomes, due to increased *de novo* autophagosome formation and due to delayed fusion of autophagosomes with late endosomes/lysosomes.[3,4]

MLN1 shares significant sequence homology with the TRP superfamily of cation channels, characterized by permeability to monovalent cations and Ca^{2+}. Mucolipin-1 is thought to function as a proton-leak channel in lysosomes, regulating lysosomal pH and hydrolytic activity.[5] MLN1 has been localized to late endosomes and lysosomes.

In addition to MLN1, mammals encode two other highly related proteins, MLN2/TRPML2 and MLN3/TRPML3. Mutations in mouse mucolipin-3 (MLN3, TRPML3) encoded by the *MCOLN3* gene, are associated with deafness and pigmentation defects in varitint-waddler mice.[6] TRPMLs have been shown to interact and to form homo- and heteromultimers resulting in a complex pattern of subcellular localization.[7]

Reagent

Supplied as a solution in 0.01 M phosphate buffered saline, pH 7.4, containing 15 mM sodium azide as a preservative.

Antibody concentration: ~2.0 mg/mL

Precautions and Disclaimer

For R&D use only. Not for drug, household, or other uses. Please consult the Safety Data Sheet for information regarding hazards and safe handling practices.

Storage/Stability

For continuous use, store at 2–8 °C for up to one month. For extended storage, freeze at –20 °C in working aliquots. Repeated freezing and thawing, or storage in "frost-free" freezers, is not recommended. If slight turbidity occurs upon prolonged storage, clarify the solution by centrifugation before use. Working dilution samples should be discarded if not used within 12 hours.

Product Profile

Immunoblotting: a working antibody concentration of 4-8 μg/mL is recommended using membrane fraction of HEK-293T expressing human Mucolipin-1.

Notes:

1. Cell lysates should **not** be boiled before loading onto SDS-PAGE, but only warmed to 60 °C for 15 minutes.
2. In order to obtain best results in various techniques and preparations, it is recommended to determine optimal working dilutions by titration.

References

1. Miedel, M.T., et al., *J. Biol. Chem.,* **281**, 12751-12759 (2006).
2. Bach, G., et al., *Eur. J. Physiol.*, **451**, 313-317 (2005).
3. Bargal, R., et al., *Nature Genet.*, **26**, 118-123 (2000).
4. Vergarajauregui, S., et al., *Hum. Mol. Genet.,* **17**, 2723-2737 (2008).
5. Soyombo, A.A., et al., *J. Biol. Chem.,* **281**, 7294-7301 (2006).
6. Di Palma, F., et al., *Proc. Natl. Acad. Sci. USA*, **99**, 14994-14999 (2002).
7. Zeevi, D.A., et al., *Biochim. Biophys. Acta,* **1772**, 851-858 (2007).

VS,GG,KAA,PHC,MAM 08/19-1