

## Product Information

### Anti-Mucolipin-3 (N-terminal)

produced in rabbit, affinity isolated antibody

Catalog Number **M7570**

#### Product Description

Anti-Mucolipin-3 (N-terminal) is produced in rabbit using as immunogen a synthetic peptide corresponding to amino acids 26-43 of human mucolipin-3 (GeneID: 55283) conjugated to KLH. This sequence is identical between human and mouse, and highly conserved in mouse and rat. The antibody is affinity-purified using the immunizing peptide immobilized on agarose.

Anti-Mucolipin-3 (N-terminal) recognizes human mucolipin-3 by immunoblotting (~75 kDa). Detection of the anti-mucolipin-3 band is specifically inhibited with the immunizing peptide.

Mucopolidosis 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).<sup>1,2</sup> MLVI 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 mucopolidoses, lysosomal hydrolase activity is not impaired in MLVI. Rather, MLVI pathophysiology has been linked to deficiency in membrane trafficking and organelle dynamics in the late endocytic pathway.<sup>2</sup> MLN1 shares significant sequence homology with the TRP superfamily of cation channels, characterized by permeability to monovalent cations and Ca<sup>2+</sup>. MLN1 is thought to function as a proton-leak channel in lysosomes, regulating lysosomal pH and hydrolytic activity.<sup>3</sup> 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 mucolipin3 (MLN3, TRPML3) encoded by the *MCOLN3* gene, are associated with deafness and pigmentation defects in varitint-waddler mice.<sup>4</sup> TRPMLs have been shown to interact to form homo- and heteromultimers resulting in a complex pattern of localization in both the lysosomal and endoplasmic reticulum compartments.<sup>5</sup>

#### 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: ~1.5 mg/mL

#### Precautions and Disclaimer

This product is for R&D use only, not for drug, household, or other uses. Please consult the Material Safety Data Sheet for information regarding hazards and safe handling practices.

#### Storage/Stability

Store at -20 °C. For continuous use, the product may be stored at 2-8 °C for up to one month. For extended storage, freeze in working aliquots at -20 °C. 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 dilutions should be discarded if not used within 12 hours.

#### Product Profile

Immunoblotting: a working antibody concentration of 0.25-0.5 µg/mL is recommended using HEK-293T cells expressing recombinant human mucolipin-3.

Note: In order to obtain the best results using various techniques and preparations, we recommend determining the optimal working dilutions by titration.

#### References

1. Bach, G., et al., *Eur. J. Physiol.*, **451**, 313-317 (2005).
2. Bargal, R., et al., *Nature Genet.*, **26**, 118-123 (2000).
3. Soyombo, A.A., et al., *J. Biol. Chem.*, **281**, 7294-7301 (2006).
4. Di Palma, F., et al., *Proc. Natl. Acad. Sci. USA*, **99**, 14994-14999 (2002).
5. Zeevi, D.A., et al., *Biochim. Biophys. Acta*, **1772**, 851-8, (2007).

MG,KAA,PHC,MAM 10/09-1