

# Rabbit Anti-mucolipin 3 antibody

SL18732R

mucolipin 3
粘Lipoprotein3抗体
MCOLN 3 ; MCOLN3 ; FLJ11006 ; FLJ36629 ; MCLN3_HUMAN ; MCOLN 3 ; MCOLN3 ; MGC71509 ; Mucolipin-3 ; TRP ML3 ; TRPML3.
Rabbit
Polyclonal
Human,Mouse,Rat,Dog,Pig,
ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100- 500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
64kDa
The cell membrane
Lyophilized or Liquid
1mg/ml
KLH conjugated synthetic peptide derived from human mucolipin 3:101-200/553
IgG
affinity purified by Protein A
0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed
This gene encodes one of members of the mucolipin cation channel proteins. Mutation studies of the highly similar protein in mice have shown that the protein is found in cochlea hair cells, and mutant mice show early-onset hearing loss and balance problems. Multiple transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Nov 2011]

### Function:

Cation channel probably playing a role in the endocytic pathway and in the control of membrane trafficking of proteins and lipids. Could play a major role in Ca2+ transport regulating lysosomal exocytosis.

#### Subunit:

Forms multimeric complexes. Interacts with PDCD6.

#### Subcellular Location: Membrane.

**Tissue Specificity:** Widely expressed in adult and fetal tissues.

#### **DISEASE:**

Mucolipidosis type IV (MLIV) [MIM:252650]: Autosomal recessive lysosomal storage disorder characterized by severe psychomotor retardation and ophthalmologic abnormalities, including corneal opacity, retinal degeneration and strabismus. Storage bodies of lipids and water-soluble substances are seen by electron microscopy in almost every cell type of the patients. Most patients are unable to speak or walk independently and reach a maximal developmental level of 1-2 years. All patients have constitutive achlorhydia associated with a secondary elevation of serum gastrin levels. MLIV may be due to a defect in sorting and/or transport along the late endocytic pathway. MLIV is found at relatively high frequency among Ashkenazi Jews.

Similarity: Belongs to the transient receptor (TC 1.A.4) family. Polycystin subfamily. MCOLN1 sub-subfamily.

## SWISS: Q8TDD5

**Gene ID:** 55283

Database links:

Entrez Gene: 55283 Human

<u>Omim: 607400</u> Human

SwissProt: Q5T4H5 Human

SwissProt: Q8TDD5 Human

Unigene: 535239 Human
<b>Important Note:</b> This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

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