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Rabbit Anti-mucolipin 1 antibody

SL18731R

Product Name:	mucolipin 1
Chinese Name:	粘Lipoprotein1抗体
Alias:	MCLN1_HUMAN ; Mcoln 1 ; Mcoln1 ; MG 2 ; MG-2 ; MG2 ; ML 4 ; ML4 ; MLIV ; MST080 ; MSTP080 ; Mucolipidin ; Mucolipin-1 ; Mucolipin1 ; TRP ML1 ; TRPML1.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Pig,Cow,Rabbit,Sheep,Monkey,Cat,
Applications:	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.
Molecular weight:	65kDa
Cellular localization:	The cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human mucolipin 1:501-580/580
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	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:	PubMed
Product Detail:	This gene encodes a memberof the transient receptor potential (TRP) cation channel gene family. The transmembrane protein localizes to intracellular vesicular membranes including lysosomes, and functions in the late endocytic pathway and in the regulation of lysosomal exocytosis. The channel is permeable to Ca(2+), Fe(2+), Na(+), K(+), and H(+), and is modulated by changes in Ca(2+) concentration. Mutations in this gene result in mucolipidosis type IV. [provided by RefSeq, Oct 2009]

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 Ca(2+) transport regulating lysosomal exocytosis.

Subcellular Location:
Cell membrane. Late endosome membrane. Lysosome membrane.Entrez Gene: 57192 Human Entrez Gene: 94178 Mouse Entrez Gene: 288371 Rat Omim: 605248 Human SwissProt: Q9GZU1 Human SwissProt: Q99J21 Mouse Unigene: 567548 Human Unigene: 631858 Human Unigene: 8356 Mouse

Tissue Specificity:
Widely expressed in adult and fetal tissues.

DISEASE:
Defects in MCOLN1 are the cause of mucolipidosis type IV (MLIV) [MIM:252650]; also known as sialolipidosis. MLIV is an 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:
Q9GZU1

Gene ID:
57192

Database links:

Entrez Gene: 57192 Human

Entrez Gene: 94178 Mouse

Entrez Gene: 288371 Rat

Omim: 605248 Human

	SwissProt: Q9GZU1 Human SwissProt: Q99J21 Mouse Unigene: 567548 Human Unigene: 631858 Human Unigene: 8356 Mouse **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.