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mucolipin 1 Rabbit pAb

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Catalog # AP57229

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q9GZU1
Predicted	Rat, Pig, Cat, Human, Mouse, Rabbit, Monkey, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	65022
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human mucolipin 1
Epitope Specificity	501-580/580
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
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
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.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	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]

Additional Information

Gene ID	57192

Other Names	Mucolipin-1, ML1, MG-2, Mucolipidin, Transient receptor potential channel mucolipin 1, TRPML1, MCOLN1 {ECO:0000303|PubMed:25720963, ECO:0000312|HGNC:HGNC:13356}
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

Name	MCOLN1 {ECO:0000303|PubMed:25720963, ECO:0000312|HGNC:HGNC:13356}
Function	Nonselective cation channel probably playing a role in the regulation of membrane trafficking events and of metal homeostasis (PubMed:11013137, PubMed:12459486, PubMed:14749347, PubMed:15336987, PubMed:18794901, PubMed:25720963, PubMed:27623384, PubMed:29019983). Acts as a Ca(2+)-permeable cation channel with inwardly rectifying activity (PubMed:25720963, PubMed:29019983). Proposed to play a major role in Ca(2+) release from late endosome and lysosome vesicles to the cytoplasm, which is important for many lysosome-dependent cellular events, including the fusion and trafficking of these organelles, exocytosis and autophagy (PubMed:11013137, PubMed:12459486, PubMed:14749347, PubMed:15336987, PubMed:25720963, PubMed:27623384, PubMed:29019983). Required for efficient uptake of large particles in macrophages in which Ca(2+) release from the lysosomes triggers lysosomal exocytosis. May also play a role in phagosome-lysosome fusion (By similarity). Involved in lactosylceramide trafficking indicative for a role in the regulation of late endocytic membrane fusion/fission events (PubMed:16978393). By mediating lysosomal Ca(2+) release is involved in regulation of mTORC1 signaling and in mTOR/TFEB-dependent lysosomal adaptation to environmental cues such as nutrient levels (PubMed:25720963, PubMed:25733853, PubMed:27787197). Seems to act as lysosomal active oxygen species (ROS) sensor involved in ROS-induced TFEB activation and autophagy (PubMed:27357649). Also functions as a Fe(2+) permeable channel in late endosomes and lysosomes (PubMed:18794901). Also permeable to Mg(2+), Na(+), K(+) and Cs(+) (By similarity). Proposed to play a role in zinc homeostasis probably implicating its association with TMEM163 (PubMed:25130899) In adaptive immunity, TRPML2 and TRPML1 may play redundant roles in the function of the specialized lysosomes of B cells (By similarity).
Cellular Location	Late endosome membrane; Multi-pass membrane protein. Lysosome membrane; Multi-pass membrane protein. Cytoplasmic vesicle membrane; Multi-pass membrane protein. Cell projection, phagocytic cup {ECO:0000250|UniProtKB:Q99J21}. Cytoplasmic vesicle, phagosome membrane {ECO:0000250|UniProtKB:Q99J21}; Multi-pass membrane protein. Cell membrane; Multi-pass membrane protein. Note=Delivery from the trans-Golgi to lysosomes seems to occur mainly in a direct intracellular manner without intermediate delivery to the plasma membrane (PubMed:16497227) Under normal conditions, restricted to intracellular compartments so that only a very minor proportion is present at the cell membrane (PubMed:12459486, PubMed:18794901, PubMed:28112729, PubMed:29019983)

Tissue Location	Widely expressed in adult and fetal tissues.

Background

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Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.