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INPP5F Rabbit pAb

Catalog # AP98876

Product Information

Application	WB, IHC-P, IHC-F, IF, E
Primary Accession	Q01968
Other Accession	Q01968
Predicted	Rat, Pig, Human, Mouse, Rabbit, Monkey, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	104205
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human INPP5F
Epitope Specificity	611-710/901
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the inositol-1,4,5-trisphosphate 5-phosphatase type II family. Contains 1 Rho-GAP domain.
SUBUNIT	Interacts with APPL1, FAM109A/SES1 and FAM109B/SES2; APPL1-binding and FAM109A-binding are mutually exclusive. Interacts with clathrin heavy chain. Interacts with several Rab GTPases, at least RAB1B, RAB5A, RAB6A, RAB8A and RAB31; these interactions may play a dual role in targeting OCRL to the specific membranes and stimulating the phosphatase activity. Interaction with RAB8A modulates OCRL recruitment to cilia.
DISEASE	Defects in OCRL are the cause of Lowe oculocerebrorenal syndrome (OCRL) [MIM:309000]. It is an X-linked multisystem disorder affecting eyes, nervous system, and kidney. It is characterized by hydrophthalmia, cataract, mental retardation, vitamin D-resistant rickets, aminoaciduria, and reduced ammonia production by the kidney. Ocular abnormalities include cataract, glaucoma, microphthalmos, and decreased visual acuity. Developmental delay, hypotonia, behavior abnormalities, and areflexia are also present. Renal tubular involvement is characterized by impaired reabsorption of bicarbonate, amino acids, and phosphate. Musculoskeletal abnormalities such as joint hypermobility, dislocated hips, and fractures may develop as consequences of renal tubular acidosis and hypophosphatemia. Cataract is the only significant manifestation in carriers and is detected by slit-lamp examination.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	The inositol polyphosphate 5-phosphatases selectively remove the phosphate from the 5-position of various phosphatidylinositols, which generate second messengers in response to extracellular signals. OCRL1 is a type II 5-phosphatase that is mutated in the oculocerebrorenal syndrome of Lowe (OCRL). OCRL is a rare X-linked disorder that is characterized in part by congenital cataracts, mental retardation, muscular hypotonia, and renal tubular dysfunction. OCRL1 has a high affinity for phosphatidylinositol 4,5-bisphosphate as well as inositol 1,4,5-trisphosphate, and inositol 1,3,4,5-tetrakisphosphate as substrates. OCRL1 is localized to the Golgi

complex and is thought to be part of the trans-Golgi network (TGN), which suggests that OCRL1 plays a role in protein sorting and trafficking within the cell.

Additional Information

Gene ID	4952
Other Names	DENT2; Dent-2; INPP5F; LOCR; NPHL2; OCRL-1; OCRL1; OCRL_HUMAN; OCRL; Inositol polyphosphate 5-phosphatase OCRL-1 (OCRL-1); Lowe oculocerebrorenal syndrome protein; Phosphatidylinositol 3, 4, 5-triphosphate 5-phosphatase; 3.1.3.36; 3.1.3.56; OCRL inositol polyphosphate-5-phosphatase; oculocerebrorenal syndrome of Lowe; Dent disease 2
Dilution	WB=1:500-2000,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	OCRL (HGNC:8108)
Synonyms	OCRL1 {ECO:0000303|Ref.2}
Function	Catalyzes the hydrolysis of the 5-position phosphate of phosphatidylinositol 4,5-bisphosphate (PtdIns(4,5)P2) and phosphatidylinositol-3,4,5-bisphosphate (PtdIns(3,4,5)P3), with the greatest catalytic activity towards PtdIns(4,5)P2 (PubMed:10764818, PubMed:15474001, PubMed:7761412, PubMed:9430698). Also, is able to hydrolyze the 5-phosphate of inositol 1,4,5-trisphosphate and of inositol 1,3,4,5-tetrakisphosphate (PubMed:15474001, PubMed:40969890, PubMed:7761412). Regulates traffic in the endosomal pathway by regulating the specific pool of phosphatidylinositol 4,5-bisphosphate that is associated with endosomes (PubMed:21971085). Involved in primary cilia assembly (PubMed:22228094, PubMed:22543976). Acts as a regulator of phagocytosis, hydrolyzing PtdIns(4,5)P2 to promote phagosome closure, through attenuation of PI3K signaling (PubMed:22072788).
Cellular Location	Cytoplasmic vesicle, phagosome membrane {ECO:0000250|UniProtKB:D3ZGS3}. Early endosome membrane. Membrane, clathrin-coated pit. Cell projection, cilium, photoreceptor outer segment. Cell projection, cilium. Cytoplasmic vesicle {ECO:0000250|UniProtKB:D3ZGS3}. Endosome. Golgi apparatus, trans-Golgi network {ECO:0000250|UniProtKB:D3ZGS3}. Lysosome Note=Also found on macropinosomes (PubMed:25869668). Colocalized with APPL1 on phagosomes (PubMed:22072788).
Tissue Location	Brain, skeletal muscle, heart, kidney, lung, placenta and fibroblasts. Expressed in the retina and the retinal pigment epithelium.

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