

Rabbit Anti-INPP5F antibody

SL11256R

Product Name:	INPP5F U
Chinese Name:	磷酸肌醇磷酸酶蛋白INPP5抗体
Alias:	Inositol polyphosphate 5 phosphatase OCRL 1; Inositol polyphosphate 5 phosphatase OCRL1; Inositol polyphosphate 5-phosphatase OCRL-1; INPP5F; LOCR; Lowe oculocerebrorenal syndrome protein; NPHL2; OCRL; OCRL_HUMAN; OCRL1; EC 3.1.3.36; Oculocerebrorenal syndrome of Lowe; Phosphatidylinositol polyphosphate 5 phosphatase.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human, Mouse, Rat, Dog, Pig, Cow, Horse, Rabbit, Sheep, GPV, Monkey,, mk
Applications:	WB=1:500-2000ELISA=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:	104kDa
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human INPP5F:611-710/901
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:	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 4,5bisphosphate as well as inositol 1,4,5-trisphosphate, and inositol 1,3,4,5tetrakisphosphate 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.

Function:

Converts phosphatidylinositol 4,5-bisphosphate to phosphatidylinositol 4-phosphate. Also converts inositol 1,4,5-trisphosphate to inositol 1,4-bisphosphate and inositol 1,3,4,5-tetrakisphosphate to inositol 1,3,4-trisphosphate. May function in lysosomal membrane trafficking by regulating the specific pool of phosphatidylinositol 4,5-bisphosphate that is associated with lysosomes.

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.

Subcellular Location:

Endosome. Also found on macropinosomes.

Tissue Specificity:

Brain, skeletal muscle, heart, kidney, lung, placenta and fibroblasts.

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.

Similarity:

Belongs to the inositol-1,4,5-trisphosphate 5-phosphatase type II family. Contains 1 Rho-GAP domain.

SWISS:

Q01968
Gene ID: 4952
Database links:
Entrez Gene: 4952 Human
<u>Omim: 300535</u> Human
<u>SwissProt: Q01968</u> Human
Unigene: 126357 Human
Unigene: 369755 Human
Unigene: 369755 Human Important Note: This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
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