

Product name:	OCRL Rabbit Polyclonal Antibody
Cat number:	ABN15095
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human OCRL. AA range:150-199
Reactivity:	Human,Mouse
Applications:	WB 1:500-1:2000,ELISA 1:5000-1:20000
Molecular Weight:	104kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

This gene encodes an inositol polyphosphate 5-phosphatase. This protein is involved in regulating membrane trafficking and is located in numerous subcellular locations including the trans-Golgi network, clathrin-coated vesicles and, endosomes and the plasma membrane. This protein may also play a role in primary cilium formation. Mutations in this gene cause oculocerebrorenal syndrome of Lowe and also Dent disease. Alternate splicing results in multiple transcript variants. [provided by RefSeq, Jan 2016],catalytic activity:1-phosphatidyl-1D-myo-inositol 4,5-bisphosphate + H(2)O = 1-phosphatidyl-1D-myo-inositol 4-phosphate + phosphate.,caution:It is uncertain whether Met-1, Met-18 or Met-20 is the initiator.,disease:Defects in OCRL are the cause of Dent disease type 2 (DD2) [MIM:300555]. DD2 is a renal disease belonging to the 'Dent disease complex', a group of disorders characterized by proximal renal tubular defect, hypercalciuria, nephrocalcinosis, and renal insufficiency. The spectrum of phenotypic features is remarkably similar in the various disorders, except for differences in the severity of bone deformities and renal impairment. Characteristic abnormalities include low-molecular-weight proteinuria and other features of Fanconi syndrome, such as glycosuria, aminoaciduria, and phosphaturia, but typically do not include proximal renal tubular acidosis. Progressive renal failure is common, as are nephrocalcinosis and kidney stones.,disease:Defects in OCRL are the cause of Lowe syndrome [MIM:309000]; also known as Lowe oculocerebrorenal syndrome. The Lowe syndrome 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.,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.,similarity:Belongs to the inositol-1,4,5-trisphosphate 5-phosphatase type II family.,similarity:Contains 1 Rho-GAP domain.,tissue specificity:Brain, skeletal muscle, heart, kidney, lung, placenta and fibroblasts.,