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Rabbit Anti-RPGRIP1L antibody

SL11946R

Product Name:	RPGRIP1L
Chinese Name:	梅克尔憩室综合征相关蛋白5抗体
Alias:	CORS 3; CORS3; Fantom; FTM; JBTS 1; JBTS 7; JBTS1; JBTS7; Joubert syndrome 1; Joubert syndrome 7; Meckel syndrome, type 1; RPGRIP1-like; Meckel syndrome, type 5; MKS 5; MKS5; NPHP 8; NPHP8; nephrocystin 8; Protein fantom; Retinitis pigmentosa GTPase regulator interacting protein 1 like; RPGR interacting protein 1 like protein; RPGRIP1 like protein; FTM_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Pig,Cow,Horse,Rabbit,Sheep,Guinea Pig,
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:	151kDa
Cellular localization:	cytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human RPGRIP1L:41-140/1315
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:	RPGRIP1L is a 1,315 amino acid protein that belongs to the RPGRIP1 family and is thought to function in programmed cell death, craniofacial development and formation of the left-right axis. Existing as two alternatively spliced isoforms that localize to the

	cytoplasm, cytoskeleton, centrosome and cilium basal body, RPGRIP1L interacts with nephrocystin-4 and is moderately expressed in brain, retina and kidney. Containing two C2 domains, RPGRIP1L is encoded by a gene that maps to human chromosome 16q12.2. Defects in the gene encoding RPGRIP1L are the cause of Joubert syndrome type 7 (JBTS7), COACH syndrome (COACHS) and Meckel syndrome type 5 (MKS5). **Function:** RPGRIP1L (retinitis pigmentosa GTPase regulator interacting protein 1 like), also known as Fantom, is a ciliary basal body protein. It is thought to play an important role in development and apoptosis. Mutations in the gene encoding RPGRIP1L are associated with Joubert's Syndrome and Meckel's Syndrome, which are characterised by a number of developmental abnormalities. **Subunit:** Interacts with NPHP4 in a complex containing NPHP1, NPHP4 and RPGRIP1L/NPHP8. Interacts with TBXA2R (via C-terminus). Interacts with IQCB1; the interaction likely requires additional interactors. Interacts with RPGR. **Subcellular Location:** Cytoplasm. Cell projection, cilium basal body. Cell projection, cilium axoneme. Centrosome. **Tissue Specificity:** Ubiquitously expressed with relatively high level of expression in hypothalamus and islet. During early development, expressed in multiple organs including brain, eye, forelimb and kidney. **DISEASE:** Note=Ciliary dysfunction leads to a broad spectrum of disorders, collectively termed ciliopathies. Overlapping clinical features include retinal degeneration, renal cystic disease, skeletal abnormalities, fibrosis of various organ, and a complex range of anatomical and functional defects of the central and peripheral nervous system. The ciliopathy range of diseases includes Meckel-Gruber syndrome, Bardet-Biedl syndrome, Joubert syndrome, nephronophtisis, Senior-Loken syndrome, and Jeune asphyxiating thoracic dystrophy among others. Single-locus allelism is insufficient to explain the variable penetrance and expressivity of such disorders, leading to the suggestion that variations across multiple sites of the ciliary proteome, including RPGRIP1L, influence the clinical outcome. Defects in RPGRIP1L are the cause of Joubert syndrome type 7 (JBTS7) [MIM:611560]. JBTS is an autosomal recessive disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermis hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease.

	Defects in RPGRIP1L are the cause of Meckel syndrome type 5 (MKS5) [MIM:611561]. MKS is an autosomal recessive disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. Defects in RPGRIP1L are a cause of COACH syndrome (COACHS) [MIM:216360]. It is a disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. **Similarity:** Belongs to the RPGRIP1 family. Contains 2 C2 domains. **SWISS:** Q68CZ1 **Gene ID:** 23322 **Database links:** Entrez Gene: 23322Human Omim: 610937Human SwissProt: Q68CZ1Human Unigene: 298382Human **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.