

Product name:	PIPK I γ Rabbit Polyclonal Antibody
Cat number:	ABN16162
Conjugate:	Unconjugated
Size:	100 μ L
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human PIP5K1C. AA range:305-354
Reactivity:	Human,Rat,Mouse
Applications:	WB 1:500-1:2000,ELISA 1:10000-1:20000
Molecular Weight:	80kDa
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:

phosphatidylinositol-4-phosphate 5-kinase type 1 gamma(PIP5K1C) Homo sapiens This locus encodes a type I phosphatidylinositol 4-phosphate 5-kinase. The encoded protein catalyzes phosphorylation of phosphatidylinositol 4-phosphate, producing phosphatidylinositol 4,5-bisphosphate. This enzyme is found at synapses and has been found to play roles in endocytosis and cell migration. Mutations at this locus have been associated with lethal congenital contractural syndrome. Alternatively spliced transcript variants encoding different isoforms have been described.[provided by RefSeq, Sep 2010],catalytic activity:ATP + 1-phosphatidyl-1D-myo-inositol 4-phosphate = ADP + 1-phosphatidyl-1D-myo-inositol 4,5-bisphosphate.,disease:Defects in PIP5K1C are the cause of lethal congenital contractural syndrome type 3 (LCCS3) [MIM:611369]; also known as multiple contractural syndrome Israeli Bedouin type B. LCCS is an autosomal recessive disorder characterized by early fetal hydrops and akinesia, the Pena-Shokeir phenotype, specific neuropathology with degeneration of anterior horn neurons and extreme skeletal muscle atrophy. LCCS3 patients present at birth with severe multiple joint contractures with severe muscle wasting and atrophy, mainly in the legs. LCCS3 can be distinguished from the original LCCS by the absence of hydrops, fractures, and multiple pterygia.,enzyme regulation:Activated by interaction with TLN2.,function:Plays a role in membrane ruffling and assembly of clathrin-coated pits at the synapse. Mediates RAC1-dependent reorganization of actin filaments (By similarity). Participates in the biosynthesis of phosphatidylinositol-4,5-bisphosphate.,similarity:Contains 1 PI5K domain.,subcellular location:Cytoplasmic, associated with the plasma membrane. Detected in focal adhesion plaques, membrane ruffles and plasma membrane invaginations.,subunit:Interacts with TLN1 and CSK (By similarity). Interacts with TLN2 and ARF6.,