SunLong Biotech Co.,LTD
Tel: 0086-571- 56623320 Fax:0086-571- 56623318
E-mail:sales@sunlongbiotech.com
www.sunlongbiotech.com

Rabbit Anti-TMEM67/Meckelin antibody

SL18756R

Product Name:	TMEM67/Meckelin
Chinese Name:	Transmembrane protein67抗体
Alias:	JBTS6; Meckel syndrome type 3 protein; MKS3; TMEM67; Transmembrane protein 67.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Cow,Horse,Rabbit,Sheep,
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:	112kDa
Cellular localization:	The cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human TMEM67/Meckelin:601-700/995
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 protein encoded by this gene localizes to the primary cilium and to the plasma membrane. The gene functions in centriole migration to the apical membrane and formation of the primary cilium. Multiple transcript variants encoding different isoforms have been found for this gene. Defects in this gene are a cause of Meckel syndrome type 3 (MKS3) and Joubert syndrome type 6 (JBTS6). [provided by RefSeq,

	Nov 2008] **Function:** Required for ciliary structure and function. Part of the tectonic-like complex which is required for tissue-specific ciliogenesis and may regulate ciliary membrane composition By similarity. Involved in centrosome migration to the apical cell surface during early ciliogenesis. Involved in the regulation of cilia length and appropriate number through the control of centrosome duplication. Required for cell branching morphology. Essential for endoplasmic reticulum-associated degradation (ERAD) of surfactant protein C (SFTPC). **Subunit:** Part of the tectonic-like complex (also named B9 complex) By similarity. Interacts with DNAJB9, DNAJC10 and mutated SFTPC. Interacts with SYNE2 during the early establishment of cell polarity. Interacts (via C-terminus) with FLNA. **Subcellular Location:** Cell membrane; Multi-pass membrane protein **Tissue Specificity:** Widely expressed in adult and fetal tissues. Expressed at higher level in spinal cord. **DISEASE:** Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early-onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. Note: The gene represented in this entry may act as a disease modifier. TMEM67 variations may influence the expression of Bardet-Biedl syndrome in patients who have causative mutations in other genes. Heterozygosity for a complex mutation in the TMEM67 gene coding for a protein with 2 in cis changes, and homozygosity for a truncating mutation of the CEP290 gene has been found in a patient with Bardet-Biedl syndrome 14. COACH syndrome (COACHS) [MIM:216360]: 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. Note: The disease is caused by mutations affecting the gene represented in this entry. Ref.15 Ref.18 Nephronophthisis 11 (NPHP11) [MIM:613550]: A disorder characterized by the association of nephronophthisis with hepatic fibrosis. Nephronophthisis is a progressive tubulo-interstitial kidney disorder histologically characterized by modifications of the tubules with thickening of the basement membrane, interstitial fibrosis and, in the

	advanced stages, medullary cysts. Typical clinical features are chronic renal failure, anemia, polyuria, polydipsia, isosthenuria, and growth retardation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. Note: The disease is caused by mutations affecting the gene represented in this entry. **SWISS:** Q5HYA8 **Gene ID:** 91147 **Database links:** Entrez Gene: 91147 Human Entrez Gene: 329795 Mouse Entrez Gene: 313067 Rat Omim: 609884 Human SwissProt: Q5HYA8 Human SwissProt: Q8BR76 Mouse SwissProt: POC152 Rat **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.