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

Rabbit Anti-MKS1 antibody

SL11513R

Product Name:	MKS1
Chinese Name:	梅克尔-格鲁伯综合征相关蛋白抗体
Alias:	MES; B8d3; BBS13; Dysencephalia splanchnocystica; FABB proteome like protein; FLJ20345; Gruber syndrome; Meckel gruber syndrome; Meckel gruber syndrome type 1; Meckel syndrome; Meckel syndrome type 1; Meckel syndrome type 1 protein; Meckel syndrome type 1 protein homolog; MKS 1; MKS; MKS1; POC12; POC12 centriolar protein homolog.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Pig,Cow,Horse,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:	64kDa
Cellular localization:	cytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human MKS1:215-300/559
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:	MKS1 is involved in centrosome migration to the apical cell surface during early ciliogenesis. It is required for ciliary structure and function, including a role in regulating length and appropriate number through modulating centrosome duplication.

It is also required for cell branching morphology.

Function:
Component of the tectonic-like complex, a complex localized at the transition zone of primary cilia and acting as a barrier that prevents diffusion of transmembrane proteins between the cilia and plasma membranes. Involved in centrosome migration to the apical cell surface during early ciliogenesis. Required for ciliary structure and function, including a role in regulating length and appropriate number through modulating centrosome duplication. Required for cell branching morphology.

Subunit:
Part of the tectonic-like complex (also named B9 complex) (By similarity). Interacts with TCTN3 and AHI1 (By similarity). Interacts with FLNA.

Subcellular Location:
Cytoplasm, cytoskeleton, cilium basal body. Cytoplasm, cytoskeleton, centrosome. Note: Localizes at the transition zone, a region between the basal body and the ciliary axoneme

DISEASE:
Defects in MKS1 are the cause of Meckel syndrome type 1 (MKS1) [MIM:249000]. MKS1 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 MKS1 are the cause of Bardet-Biedl syndrome type 13 (BBS13) [MIM:209900]. Bardet-Biedl syndrome (BBS) is a genetically heterogeneous, autosomal recessive disorder 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. A relatively high incidence of BBS is found in the mixed Arab populations of Kuwait and in Bedouin tribes throughout the Middle East, most likely due to the high rate of consaguinity in these populations and a founder effect.

Similarity:
Contains 1 B9 domain.

SWISS:
Q9NXB0

Gene ID:
54903

Database links:

Entrez Gene: 54903Human

Entrez Gene: 380718Mouse

Entrez Gene: 287612Rat

Omim: 609883Human

SwissProt: Q9NXB0Human

SwissProt: Q5SW45Mouse

SwissProt: Q499Q5Rat

Unigene: 408843Human

Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.