巴尔得-别德尔综合征相关蛋白 9 抗体

产品货号： mlR11511

英文名称： BBS9

中文名称： 巴尔得-别德尔综合征相关蛋白 9 抗体

别　　名： B1 antibody; Bardet Biedl syndrome 9; Bardet-Biedl syndrome 9 protein; bbs9; C18 antibody D1 antibody MGC118917; 1 gene protein; Protein PTHB1; PTH-responsive osteosarcoma B1 protein; PTHB1; PTHB1_HUMAN.

研究领域： 肿瘤　神经生物学　信号转导　生长因子和激素　内分泌病

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Dog, Pig, Horse, Rabbit, Sheep,

产品应用： ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500 （石蜡切片需做抗原修复）
not yet tested in other applications.
optimal dilutions/concentrations should be determined by the end user.

分 子 量： 99kDa

细胞定位： 细胞浆 细胞膜

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human BBS9:244-320/887

亚　　型： IgG

纯化方法： affinity purified by Protein A

储 存 液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： 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

产品介绍 ： BBS9 is an 887 amino acid protein that localizes to both the cytoplasm and the centrosome and exists as six alternatively spliced isoforms. Expressed in a wide variety of tissues, including liver, lung, heart, brain and skeletal muscle, BBS9 functions as a component of the multi-protein BBSome complex which is required for ciliogenesis and is regulated by GDP/GTP exchange factors. Defects in the gene encoding BBS9 are associated with the pathogenesis of Bardet-Biedl syndrome type 9 (BBS9), an autosomal recessive disorder that is characterized by severe pigmentary retinopathy, early onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Additionally, chromosomal aberrations involving the BBS9 gene may play a role in the formation of Wilms tumor 5 (WT5).

Function:

The BBSome complex is required for ciliogenesis but is dispensable for centriolar satellite function. This ciliogenic function is mediated in part by the Rab8 GDP/GTP exchange factor, which localizes to the basal body and contacts the BBSome. Rab8(GTP) enters the primary cilium and promotes extension of the ciliary membrane. Firstly the BBSome associates with the ciliary membrane and binds to RAB3IP/Rabin8, the guanosyl exchange factor (GEF) for Rab8 and then the Rab8-GTP localizes to the cilium and promotes docking and fusion of carrier vesicles to the base of the ciliary membrane.

Subunit:

Part of BBSome complex, that contains BBS1, BBS2, BBS4, BBS5, BBS7, BBS8, BBS9 and BBIP10. The BBSome complex binds to PCM1 and tubulin.

Subcellular Location:

Cytoplasm, cytoskeleton, centrosome. Cell projection, cilium membrane. Cytoplasm. Note=Localizes to nonmembranous centriolar satellites in the cytoplasm.

Tissue Specificity:

Widely expressed. Expressed in adult heart, skeletal muscle, lung, liver, kidney, placenta and brain, and in fetal kidney, lung, liver and brain.

DISEASE:

Defects in BBS9 are a cause of Bardet-Biedl syndrome type 9 (BBS9) [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.

SWISS:

Q3SYG4

Gene ID:

27241

Important Note:

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

产品图片

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