巴尔得-别德尔综合征相关蛋白 4 抗体

产品货号： mlR11508

英文名称： BBS4

中文名称： 巴尔得-别德尔综合征相关蛋白 4 抗体

别　　名： Bardet Biedl syndrome 4 protein; Bardet-Biedl syndrome 4 protein; Bbs4; BBS4_HUMAN.

研究领域： 肿瘤　细胞生物　神经生物学　内分泌病

抗体来源： Rabbit

克隆类型： Polyclonal

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

产品应用： WB=1:500-2000 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： 58kDa

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 BBS4:431-519/519

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： Bardet-Biedl syndrome (BBS) is a pleiotropic genetic disorder characterized by obesity, photoreceptor degeneration, polydactyly, hypogenitalism, renal abnormalities, and developmental delay. Other associated clinical findings in BBS patients include diabetes, hypertension, and congenital heart defects. BBS is a heterogeneous disorder; BBS genes map to eight genetic loci and encode eight proteins, BBS1-BBS8. Five BBS genes encode basal body or cilia proteins, suggesting that BBS is a ciliary dysfunction disorder. BBS4 is expressed in the olfactory epithelium and localizes to the centriolar satellites of centrosomes and basal bodies of primary cilia. BBS4 regulates the p150 subunit of the dynein transport machinery (DCTN1) to attract pericentriolar material-1 protein (PCM1) and its associated components to the satellites. Loss of BBS4 is correlated with obesity caused by abnormal lipid profiles, liver dysfunction, elevated insulin, and abnormal leptin levels.

Function:

May be required for the dynein-mediated transport of pericentriolar proteins to the centrosome. Required for microtubule anchoring at the centrosome but not for microtubule nucleation. 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 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. Interacts with DCTN1. Interacts with CCDC28B.

Subcellular Location:

Cytoplasm, cytoskeleton, centrosome. Cytoplasm, cytoskeleton. Cell projection, cilium membrane. Cytoplasm. Note=Localizes to the pericentriolar region throughout the cell cycle. Centrosomal localization requires dynein. Localizes to nonmembranous centriolar satellites in the cytoplasm.

Tissue Specificity:

Ubiquitously expressed. The highest level of expression is found in the kidney.

DISEASE:

Defects in BBS4 are the cause of Bardet-Biedl syndrome type 4 (BBS4) [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:

Belongs to the BBS4 family.

Contains 10 TPR repeats.

SWISS:

Q96RK4

Gene ID:

585

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