巴德-毕德氏综合征蛋白 BBS5 抗体

产品货号： mlR9438

英文名称： BBS5

中文名称： 巴德-毕德氏综合征蛋白 BBS5 抗体

别　　名： Bardet Biedl syndrome 5 protein; BBS 5; BBS-5.

研究领域： 肿瘤　细胞生物　神经生物学

抗体来源： Rabbit

克隆类型： Polyclonal

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

产品应用： ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 IF=1:50-200 （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： 38kDa

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 BBS5:241-341/341

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. BBS5 localizes to ciliary basal bodies and is a member of the basal body/flagellar proteome. It plays a role in flagellar and basal body assembly and function. A mutation or loss of BBS5 may be correlated with photoreceptor degeneration

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. Binds to phosphoinositides.

Subcellular Location:

Cell projection, cilium membrane. Cytoplasm. Cytoplasm, cytoskeleton, cilium basal body.

DISEASE:

Defects in BBS5 are a cause of Bardet-Biedl syndrome type 5 (BBS5) [MIM:209900]. Bardet-Biedl syndrome (BBS) is a genetically heterogeneous 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 BBS5 family.

SWISS:

Q8N3I7

Gene ID:

129880

Important Note:

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

BBS 蛋白是一类研究早期儿童肥胖综合症有关的其中一种。巴尔得-别德尔综合征（Bardet-Biedl syndrome，BBS）的特征为不同程度的肥胖、智力延迟、色素视网膜病变、多指和肾脏异常。

产品图片