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BBS5 Rabbit pAb

Catalog # AP98630

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q8N3I7
Other Accession	Q8N3I7
Reactivity	Mouse
Predicted	Rat, Pig, Human, Dog, Horse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	38755
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human BBS5
Epitope Specificity	241-341/341
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the BBS5 family.
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.
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.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	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

Additional Information

Gene ID	129880

Other Names	1700049I01Rik; 2700023J09Rik; BBS5_HUMAN; BBS5; Bardet-Biedl syndrome 5 protein; Bardet-Biedl syndrome 5
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

Name	BBS5
Function	The BBSome complex is thought to function as a coat complex required for sorting of specific membrane proteins to the primary cilia. 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. The BBSome complex, together with the LTZL1, controls SMO ciliary trafficking and contributes to the sonic hedgehog (SHH) pathway regulation. Required for BBSome complex ciliary localization but not for the proper complex assembly.
Cellular Location	Cell projection, cilium membrane. Cytoplasm. Cytoplasm, cytoskeleton, cilium basal body. Cytoplasm, cytoskeleton, microtubule organizing center, centrosome, centriolar satellite. Note=Localizes to basal bodies.

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