

BBS7 Rabbit pAb

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Catalog # AP54529

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	Q8IWZ6
Predicted	Rat, Pig, Dog, Human, Mouse, Rabbit, Chicken, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	80353
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human BBS7
Epitope Specificity	551-620/715
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Cell projection, cilium membrane. Cytoplasm
DISEASE	Note=Ciliary dysfunction leads to a broad spectrum of disorders, collectively termed ciliopathies. Overlapping clinical features include retinal degeneration, renal cystic disease, skeletal abnormalities, fibrosis of various organ, and a complex range of anatomical and functional defects of the central and peripheral nervous system. The ciliopathy range of diseases includes Meckel-Gruber syndrome, Bardet-Biedl syndrome, Joubert syndrome, nephronophthisis, Senior-Loken syndrome, and Jeune asphyxiating thoracic dystrophy among others. Single-locus allelism is insufficient to explain the variable penetrance and expressivity of such disorders, leading to the suggestion that variations across multiple sites of the ciliary proteome, including BBS7, influence the clinical outcome. Defects in BBS7 are a cause of Bardet-Biedl syndrome type 7 (BBS7) [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 consanguinity in these populations and a founder effect. Inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for disease manifestation in some cases (triallelic inheritance).
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. BBS patients also have an increased risk of developing diabetes, hypertension, and congenital heart defects. BBS is a heterogeneous disorder mapping to eight genetic loci and encoding eight proteins, BBS1-BBS8. Five BBS proteins encode basal body or cilia proteins,

suggesting that BBS is a ciliary dysfunction disorder. BBS2 contains two overlapping genes: BBS2L1 and BBS2L2. BBSL1 was re-named BBS7, whereas BBS2L2 independently functions as BBS1. BBS7 contains 672 amino acids and is expressed at low to moderate levels in most human tissues.

Additional Information

Gene ID	55212
Other Names	BBSome complex member BBS7, BBS2-like protein 1, Bardet-Biedl syndrome 7 protein, BBS7, BBS2L1
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	BBS7
Synonyms	BBS2L1
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 proper BBSome complex assembly and its ciliary localization.
Cellular Location	Cell projection, cilium membrane. Cytoplasm. Cytoplasm, cytoskeleton, microtubule organizing center, centrosome, centriolar satellite. Cytoplasm, cytoskeleton, cilium basal body {ECO:0000250 UniProtKB:Q8K2G4}
Tissue Location	Isoform 2 is ubiquitously expressed. Isoform 1 is expressed in retina, lung, liver, testis, ovary, prostate, small intestine, liver, brain, heart and pancreas

Background

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Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.