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Rabbit Anti-BBS7 antibody

SL11509R

Product Name:	BBS7
Chinese Name:	巴尔得-别德尔综合征相关蛋白7抗体
Alias:	Bardet-Biedl syndrome 7; Bardet-Biedl syndrome 7 protein; BBS2-like 1; BBS7_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Dog,Pig,Cow,Horse,Rabbit,Sheep,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	80kDa
Cellular localization:	cytoplasmicThe cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human BBS7:551-620/715
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	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
Product Detail:	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 funcitons as BBS1. BBS7 contains 672 amino acids and is expressed at low to moderate levels in most human tissues.

Function:
BBS7 is a widely expressed protein with similarity to BBS2. Defects in BBS7 are a cause of Bardet-Biedl syndrome type 7 (BBS7) which is a genetically heterogeneous disorder characterized by usually severe pigmentary retinopathy, early onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. The encoded protein may play a role in eye, limb, cardiac and reproductive system development. Two transcript variants encoding distinct isoforms have been identified for this gene.

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 BBS2 (via C-terminus). Interacts with CCDC28B.

Subcellular Location:
Cell projection, cilium membrane. Cytoplasm

Tissue Specificity:
soform 2 is ubiquitously expressed. Isoform 1 is expressed in retina, lung, liver, testis, ovary, prostate, small intestine, liver, brain, heart and pancreas.

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, nephronophtisis, 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 consaguinity 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).

SWISS:
Q8IWZ6

Gene ID:
55212

Database links:

Entrez Gene: 55212Human

Entrez Gene: 71492Mouse

Entrez Gene: 361930Rat

Omim: 607590Human

SwissProt: Q8IWZ6Human

SwissProt: Q8K2G4Mouse

Unigene: 591694Human

Unigene: 286187Mouse

Unigene: 28442Rat

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。