SunLong Biotech Co.,LTD
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[Primary Antibody]

Rabbit Anti-BBS7 antibody

Catalog Number: SL11509R

FOR RESEARCH USE ONLY

[DATASHEET]

Host:	Rabbit
Isotype:	IgG
Clonality:	Polyclonal
Target:	BBS7
Immunogen:	KLH conjugated synthetic peptide derived from human BBS7: 551-620/715
Purification:	affinity purified by Protein A
Concentration:	1mg/ml
Storage:	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol. Shipped at 4℃. Store at -20 °C for one year. Avoid repeated freeze/thaw cycles.
Applications:	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 （Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Reactivity:	(predicted: Human, Mouse, Rat, Chicken, Dog, Pig, Cow, Horse, Rabbit, Sheep)
Predicted MW.:	80kDa
Subcellular Location:	cytoplasmic The cell membrane
Background:	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

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	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). **Database links:** **Entrez Gene:** 55212 Human

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	Entrez Gene: 71492 Mouse **Entrez Gene:** 361930 Rat Omim: 607590 Human **SwissProt:** Q8IWZ6 Human **SwissProt:** Q8K2G4 Mouse **Unigene:** 591694 Human **Unigene:** 286187 Mouse **Unigene:** 28442 Rat BBS proteins are one of the types associated with research on early childhood obesity syndrome. Bardet-Biedl syndrome (BBS) is characterized by varying degrees of obesity, intellectual delay, pigmentary retinopathy, polydactyly, and renal abnormalities.

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