



BBS7 rabbit pAb

Catalog No	BYab-11680
Isotype	IgG
Reactivity	Human; Mouse
Applications	WB
Gene Name	BBS7 BBS2L1
Protein Name	BBS7
Immunogen	Synthesized peptide derived from human BBS7 AA range: 85-135
Specificity	This antibody detects endogenous levels of BBS7 at Human/Mouse
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.
Source	Polyclonal, Rabbit,IgG
Purification	The antibody was affinity-purified from rabbit serum by affinity-chromatography using specific immunogen.
Dilution	WB 1: 500-2000
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	
Observed Band	
Cell Pathway	Cell projection, cilium membrane . Cytoplasm . Cytoplasm, cytoskeleton, microtubule organizing center, centrosome, centriolar satellite . Cytoplasm, cytoskeleton, cilium basal body .
Tissue Specificity	Isoform 2 is ubiquitously expressed. Isoform 1 is expressed in retina, lung, liver, testis, ovary, prostate, small intestine, liver, brain, heart and pancreas.
Function	disease: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 obseity, polydactyly, bypogenitalism, renal malformation and mental retardation.

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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

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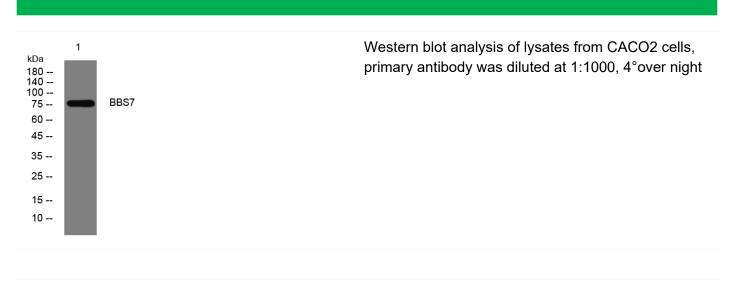


attention	This product can be used in immunological reaction related experiments. For
matters needing	Avoid repeated freezing and thawing!
Background	This gene encodes one of eight proteins that form the BBSome complex containing BBS1, BBS2, BBS4, BBS5, BBS7, BBS8, BBS9 and BBIP10. The BBSome complex is believed to recruit Rab8(GTP) to the primary cilium and promote ciliogenesis. The BBSome complex assembly is mediated by a complex composed of three chaperonin-like BBS proteins (BBS6, BBS10, and BBS12) and CCT/TRiC family chaperonins. Mutations in this gene are implicated in Bardet-Biedl syndrome, a genetic disorder whose symptoms include obesity, retinal degeneration, polydactyly and nephropathy; however, mutations in this gene and the BBS8 gene are thought to play a minor role and mutations in chaperonin-like BBS genes are found to be a major contributor to disease development in a multiethnic Bardet-Biedl syndrome patient population. Two transcript variants encoding distinct isoforms have been identified for this gene.[provided by RefSeq, Oct 2014],
	inheritance).,function:The BBSome complex is required for ciliogenesis but is dispensable for centr

Usage suggestions

This product can be used in immunological reaction related experiments. For more information, please consult technical personnel.

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