BBS4 Antibody – Rabbit Polyclonal

PRODUCT OVERVIEW

Product Name	BBS4, Polyclonal Antibody
Full Product Name	BBS4 (Center) Rabbit pAb
Product Gene Name	anti-BBS4 antibody
Product Synonym Names	Bardet-Biedl syndrome 4 protein; BBS4
Clonality	Polyclonal
Isotype	Ig
Clone Number	RB14682
Host	Rabbit
Species Reactivity	Human
Form/Format	Supplied in PBS with 0.09% (W/V) Sodium Azide. This antibody is prepared by Saturated Ammonium Sulfate (SAS) precipitation followed by dialysis against PBS.
Purity/Purification	Affinity Purified
OMIM	gene 615982
3D Structure	ModBase 3D Structure for Q96RK4
Immunogen	This BBS4 antibody is generated from rabbits immunized with a KLH conjugated synthetic peptide between 337-369 amino acids from the central region of human BBS4.
Cellular Localization	Cell Membrane, Cell Projection, Cilium, Cytoplasm, Cytoskeleton, Membrane
ISO Certification	Manufactured in an ISO 9001:2015 Certified Laboratory.
Applications Tested/Suitable for anti-BBS4 antibody	Western Blot (WB)
Application Notes for anti-BBS4 antibody	WB: 1:1000
Related Product Information for anti-BBS4 antibody	MBS8555016 is an antibody targeting BBS4. Also known as BBS4 (Center) Rabbit pAb. Reactive in Human. Host: Rabbit.
Other Notes	Small volumes of anti-BBS4 antibody vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply.

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for BBS4. It may not necessarily be applicable to this product.

NCBI GI #	160359000
NCBI GeneID	585
NCBI Accession #	Q96RK4.2
UniProt Primary Accession #	Q96RK4
UniProt Related Accession #	Q96RK4

NCBI Official Full Name	Bardet-Biedl syndrome 4 protein
NCBI Official Synonym Full Names	Bardet-Biedl syndrome 4
NCBI Official Symbol	BBS4
UniProt Protein Name	Bardet-Biedl syndrome 4 protein
Protein Family	Bardet-Biedl syndrome 4 protein
UniProt Gene Name	BBS4
UniProt Entry Name	BBS4_HUMAN
NCBI Protein Information	Bardet-Biedl syndrome 4 protein
NCBI Summary for BBS4	This gene is a member of the Bardet-Biedl syndrome (BBS) gene family. Bardet-Biedl syndrome is an autosomal recessive disorder characterized by severe pigmentary retinopathy, obesity, polydactyly, renal malformation and cognitive disability. The proteins encoded by BBS gene family members are structurally diverse. The similar phenotypes exhibited by mutations in BBS gene family members are likely due to the protein's shared roles in cilia formation and function. Many BBS proteins localize to the basal bodies, ciliary axonemes, and pericentriolar regions of cells. BBS proteins may also be involved in intracellular trafficking via microtubule-related transport. The protein encoded by this gene has sequence similarity to O-linked N-acetylglucosamine (O-GlcNAc) transferases in plants and archaebacteria and in human forms a multi-protein "BBSome" complex with seven other BBS proteins. Alternate splicing results in multiple transcript variants. [provided by RefSeq, Mar 2016]
UniProt Comments for BBS4	BBS4: May be required for the dynein-mediated transport of pericentriolar proteins to the centrosome. Required for microtubule anchoring at the centrosome but not for microtubule nucleation. 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. Defects in BBS4 are the cause of Bardet-Biedl syndrome type 4 (BBS4). Bardet-Biedl syndrome (BBS) is a genetically heterogeneous, autosomal recessive 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. Belongs to the BBS4 family. 2 isoforms of the human protein are produced by alternative splicing. Protein type: Cell development/differentiation; Adaptor/scaffold; Microtubule-binding Chromosomal Location of Human Ortholog: 15q22.3-q23 Cellular Component: centriole; centrosome; pericentriolar material; nonmotile primary cilium; cytosol; cilium Molecular Function: protein binding; beta-tubulin binding; microtubule motor activity; alpha-tubulin binding Biological Process: fat cell differentiation; retinal rod cell development; regulation of lipid metabolic process; metabolic process; photoreceptor cell maintenance; positive regulation of multicellular organism growth; retinal homeostasis; protein transport; sensory processing; intracellular transport; visual perception; cytokinesis after mitosis; sensory cilium biogenesis; centrosome organization and biogenesis; dendrite development; brain morphogenesis; cilium biogenesis; heart looping; maintenance of protein localization in nucleus; positive regulation of flagellum biogenesis; striatum development; negative regulation of systemic arterial blood pressure; organelle organization and biogenesis; hippocampus development; sensory perception of smell; microtubule cytoskeleton organization and biogenesis; regulation of cytokinesis; melanosome transport; protein localization in organelle; adult behavior; neural tube closure; cerebral cortex development; spermatid development Disease: Bardet-biedl Syndrome 4; Bardet-biedl Syndrome 1
Research Articles on BBS4	**1.** a novel nonsense mutation in BBS4 gene in a Chinese family with Bardet-Biedl syndrome. This homozygous mutation was predicted to completely abolish the synthesis of the BBS4 protein; a rare heterozygous missense SNP in BBS10 gene was also detected

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