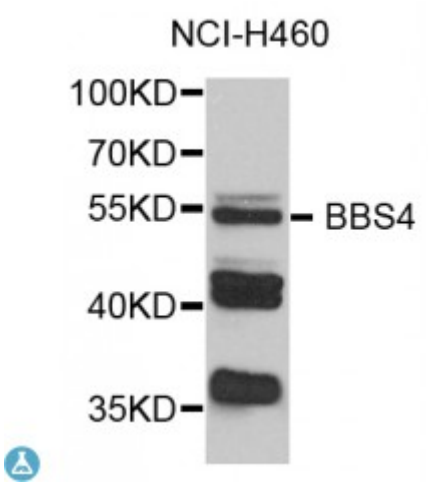
Anti-BBS4 Antibody



Description	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 mental retardation. 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.

Model	STJ117805
Host	Rabbit
Reactivity	Human
Applications	IF, WB
Immunogen	Recombinant fusion protein containing a sequence corresponding to amino acids 350-519 of human BBS4 (NP_149017.2).
Gene ID	585
Gene Symbol	BBS4
Dilution range	WB 1:500 - 1:2000 IF 1:50 - 1:200

Tissue Specificity	Ubiquitously expressed, The highest level of expression is found in the kidney
Purification	Affinity purification
Note	For Research Use Only (RUO).
Protein Name	Bardet-Biedl syndrome 4 protein
Molecular Weight	58.282 kDa
Clonality	Polyclonal
Conjugation	Unconjugated
Isotype	IgG
Formulation	PBS with 0.02% sodium azide, 50% glycerol, pH7.3.
Storage Instruction	Store at -20C. Avoid freeze / thaw cycles.
Database Links	HGNC:969OMIM:600374Reactome:R-HSA-5620922
Alternative Names	Bardet-Biedl syndrome 4 protein
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, Required for microtubule anchoring at the centrosome but not for microtubule nucleation, May be required for the dynein-mediated transport of pericentriolar proteins to the centrosome,
Cellular Localization	Cytoplasm, cytoskeleton, microtubule organizing center, centrosome,