10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

# TTC8 Rabbit pAb

Catalog # AP98257

## Product Information

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| **Application** | WB, IHC-P, IHC-F, IF |
| **Primary Accession** | Q8TAM2 |
| **Other Accession** | Q8TAM2 |
| **Reactivity** | Human, Mouse |
| **Predicted** | Rat, Pig, Rabbit, Chicken, Dog, Horse, Sheep |
| **Host** | Rabbit |
| **Clonality** | Polyclonal |
| **Calculated MW** | 61534 |
| **Physical State** | Liquid |
| **Immunogen** | KLH conjugated synthetic peptide derived from human BBS8 |
| **Epitope Specificity** | 251-330/541 |
| **Isotype** | IgG |
| **Purity** | affinity purified by Protein A |
| **Buffer** | 0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol. |
| **SIMILARITY** | Contains 8 TPR repeats. |
| **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 PCM1. Interacts with CCDC28B. |
| **DISEASE** | Defects in TTC8 are the cause of retinitis pigmentosa type 51 (RP51) [MIM:613464]. It is a retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. Defects in TTC8 are the cause of Bardet-Biedl syndrome type 8 (BBS8) [MIM:209900]. 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. |
| **Important Note** | This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications. |
| **Background Descriptions** | 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.

## Additional Information

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| **Gene ID** | 123016 |
| **Other Names** | BBS8; RP51; 0610012F22Rik; TTC8_HUMAN; TTC8; TPR repeat protein 8; Bardet-Biedl syndrome 8 protein; TTC8_MOUSE; Bardet-Biedl syndrome 8 protein homolog; tetratricopeptide repeat domain 8 |
| **Dilution** | WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500,Flow-Cyt=0.2 ug/test |
| **Storage** | Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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. |

## Protein Information

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| **Name** | TTC8 |
| **Synonyms** | BBS8 |
| **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. |
| **Cellular Location** | Cytoplasm, cytoskeleton, microtubule organizing center, centrosome. Cell projection, cilium membrane. Cytoplasm. Cytoplasm, cytoskeleton, microtubule organizing center, centrosome, centriolar satellite. Cell projection, cilium {ECO:0000250\|UniProtKB:Q8VD72} |
| **Tissue Location** | Widely expressed. |

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