

# BBS1 (S-19): sc-49791

## 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 genes encode basal body or cilia proteins, suggesting that BBS is a ciliary dysfunction disorder. BBS1 is the protein most commonly involved in Bardet-Biedl syndrome. The BBS1 gene is ubiquitously expressed, with highest abundance in fetal tissues, testes, retina, and adipose tissue. BBS1 is highly conserved in mammals and is inherited in an autosomal recessive manner. Missense mutations in the BBS1 gene account for approximately 80% of all BBS1 mutations.

## REFERENCES

1. Badano, J.L., et al. 2003. Heterozygous mutations in BBS1, BBS2 and BBS6 have a potential epistatic effect on Bardet-Biedl patients with two mutations at a second BBS locus. *Hum. Mol. Genet.* 12: 1651-1659.
2. Mykytyn, K., et al. 2003. Evaluation of complex locus (BBS1). *Am. J. Hum. Genet.* 72: 429-437.
3. Dollfus, H., et al. 2005. Update on Bardet-Biedl syndrome. *J. Fr. Ophthalmol.* 28: 106-112.
4. Fan, Y., et al. 2005. Linkage disequilibrium mapping in the Newfoundland population: a re-evaluation of the refinement of the Bardet-Biedl syndrome 1 critical interval. *Hum. Genet.* 116: 62-71.
5. Hartmann, T.B., et al. 2005. SEREX identification of new tumor antigens linked to melanoma-associated retinopathy. *Int. J. Cancer* 114: 88-93.
6. Heon, E., et al. 2005. Ocular phenotypes of three genetic variants of Bardet-Biedl syndrome. *Am. J. Med. Genet. A* 132: 283-287.
7. Hichri, H., et al. 2005. Testing for triallelism: analysis of six BBS genes in a Bardet-Biedl syndrome family cohort. *Eur. J. Hum. Genet.* 13: 607-616.

## CHROMOSOMAL LOCATION

Genetic locus: BBS1 (human) mapping to 11q13.2; Bbs1 (mouse) mapping to 19 A.

## SOURCE

BBS1 (S-19) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of BBS1 of human origin.

## PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-49791 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

Available as TransCruz reagent for gel supershift and ChIP applications, sc-49791 X, 200 µg/0.1 ml.

## APPLICATIONS

BBS1 (S-19) is recommended for detection of BBS1 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500), immunohistochemistry (including paraffin-embedded sections) (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

BBS1 (S-19) is also recommended for detection of BBS1 in additional species, including canine.

Suitable for use as control antibody for BBS1 siRNA (h): sc-60249, BBS1 siRNA (m): sc-60250, BBS1 shRNA Plasmid (h): sc-60249-SH, BBS1 shRNA Plasmid (m): sc-60250-SH, BBS1 shRNA (h) Lentiviral Particles: sc-60249-V and BBS1 shRNA (m) Lentiviral Particles: sc-60250-V.

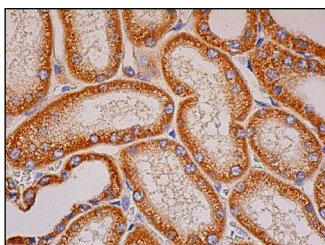
BBS1 (S-19) X TransCruz antibody is recommended for gel supershift and ChIP applications.

Molecular Weight of BBS1: 65 kDa.

## RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941. 3) Immunohistochemistry: use ImmunoCruz™: sc-2053 or ABC: sc-2023 goat IgG Staining Systems.

## DATA



BBS1 (S-19): sc-49791. Immunoperoxidase staining of formalin fixed, paraffin-embedded human kidney tissue showing cytoplasmic staining of cells in tubules.

## STORAGE

Store at 4° C, **\*\*DO NOT FREEZE\*\***. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

## RESEARCH USE

For research use only, not for use in diagnostic procedures.