

# BBS7 (D-20): sc-49803

## 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; BBS genes map to eight genetic loci and encode eight proteins, BBS1-BBS8. Five BBS genes encode basal body or cilia proteins, suggesting that BBS is a ciliary dysfunction disorder. The BBS2 gene contains two overlapping genes: BBS2L1 and BBS2L2. BBSL1 was re-named BBS7, whereas BBS2L2 independently functions as BBS1. BBS7 contains 672 amino acids and is expressed at low to moderate levels in most human tissues.

## REFERENCES

1. Myktyyn, K., et al. 2002. Identification of the gene (BBS1) most commonly involved in Bardet-Biedl syndrome, a complex human obesity syndrome. *Nat. Genet.* 31: 435-438.
2. Online Mendelian Inheritance in Man, OMIM™. 2002. Johns Hopkins University, Baltimore, MD. MIM Number: 607590. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
3. Badano, J.L., et al. 2003. Identification of a novel Bardet-Biedl syndrome protein, BBS7, that shares structural features with BBS1 and BBS2. *Am. J. Hum. Genet.* 72: 650-658.
4. Heon, E., et al. 2005. Ocular phenotypes of three genetic variants of Bardet-Biedl syndrome. *Am. J. Med. Genet. A* 132A: 283-287.
5. Nakane, T., et al. 2005. No evidence for triallelic inheritance of MKKS/BBS loci in Amish Mckusick-Kaufman syndrome. *Am. J. Med. Genet. A* 138A: 32-34.
6. 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.
7. Dollfus, H., et al. 2005. Update on Bardet-Biedl syndrome. *J. Fr. Ophthalmol.* 28: 106-112.

## CHROMOSOMAL LOCATION

Genetic locus: BBS7 (human) mapping to 4q27; Bbs7 (mouse) mapping to 3 B.

## SOURCE

BBS7 (D-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of BBS7 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-49803 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-49803 X, 200 µg/0.1 ml.

## APPLICATIONS

BBS7 (D-20) is recommended for detection of BBS7 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

BBS7 (D-20) is also recommended for detection of BBS7 in additional species, including equine, canine, bovine and porcine.

Suitable for use as control antibody for BBS7 siRNA (h): sc-60259, BBS7 siRNA (m): sc-60260, BBS7 shRNA Plasmid (h): sc-60259-SH, BBS7 shRNA Plasmid (m): sc-60260-SH, BBS7 shRNA (h) Lentiviral Particles: sc-60259-V and BBS7 shRNA (m) Lentiviral Particles: sc-60260-V.

BBS7 (D-20) X TransCruz antibody is recommended for Gel Supershift and ChIP applications.

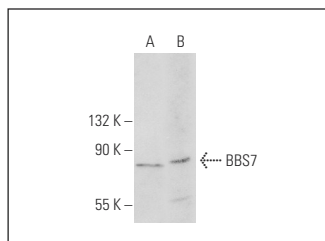
Molecular Weight of BBS7: 80 kDa.

Positive Controls: A-431 whole cell lysate: sc-2201 or mouse heart extract: sc-2254.

## 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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) 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.

## DATA



BBS7 (D-20): sc-49803. Western blot analysis of BBS7 expression in A-431 whole cell lysate (A) and mouse heart tissue extract (B).

## 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.