

## BBS3 (C-13): sc-49794

### BACKGROUND

Bardet-Biedl syndrome (BBS) is a pleiotropic genetic disorder characterized by obesity, photoreceptor degeneration, polydactyly, hypogonadism, 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. Bardet-Biedl syndrome-3 (BBS3) results from a homozygous C-to-T transition in exon 7 of the ARL6 gene, resulting in an Arg 122 to Ter mutation with a premature truncation of the protein from 186 to 121 amino acids. Heterozygosity in a mutation of the BBS3 gene modifies the expression of the Met 390 to Arg mutation in the BBS1 gene.

### REFERENCES

1. Bruford, E.A., et al. 1997. Linkage mapping in 29 Bardet-Biedl syndrome families confirms loci in chromosomal regions 11q13, 15q22.3-q23, and 16q21. *Genomics* 411: 93-99.
2. Young, T.L., et al. 1998. Canadian Bardet-Biedl syndrome family reduces the critical region of BBS3 (3p) and presents with a variable phenotype. *Am. J. Med. Genet.* 78: 461-467.
3. Ghadami, M., et al. 2000. Bardet-Biedl syndrome type 3 in an Iranian family: clinical study and confirmation of disease localization. *Am. J. Med. Genet.* 94: 433-437.

### CHROMOSOMAL LOCATION

Genetic locus: ARL6 (human) mapping to 3q11.2; Arl6 (mouse) mapping to 16 C1.3.

### SOURCE

BBS3 (C-13) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the C-terminus of BBS3 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. Also available as TransCruz reagent for Gel Supershift and ChIP applications, sc-49794 X, 200 µg/0.1 ml.

Blocking peptide available for competition studies, sc-49794 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

### STORAGE

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

### PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) or our catalog for detailed protocols and support products.

### APPLICATIONS

BBS3 (C-13) is recommended for detection of BBS3 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). BBS3 (C-13) is also recommended for detection of BBS3 in additional species, including canine.

Suitable for use as control antibody for BBS3 siRNA (h): sc-60253, BBS3 siRNA (m): sc-60254, BBS3 shRNA Plasmid (h): sc-60253-SH, BBS3 shRNA Plasmid (m): sc-60254-SH, BBS3 shRNA (h) Lentiviral Particles: sc-60253-V and BBS3 shRNA (m) Lentiviral Particles: sc-60254-V.

BBS3 (C-13) X TransCruz antibody is recommended for gel supershift and ChIP applications.

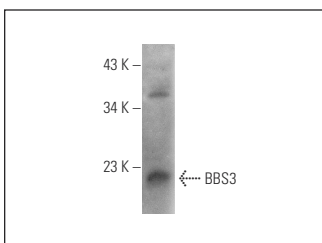
Molecular Weight of BBS3: 21 kDa.

Positive Controls: Hep G2 cell lysate: sc-2227.

### 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



BBS3 (C-13): sc-49794. Western blot analysis of BBS3 expression in 293T whole cell lysate.

### RESEARCH USE

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



Try **BBS3 (C-5): sc-390021**, our highly recommended monoclonal alternative to BBS3 (C-13).