SANTA CRUZ BIOTECHNOLOGY, INC.

BBS3 (C-5): sc-390021

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. Bardet-Biedl syndrome-3 (BBS3) results from a homozygous C-to-T transition in exon 7 of the ARL6 gene, resulting in an arg122-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 met390-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 41: 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.
4. Online Mendelian Inheritance in Man, OMIM™. 2002. Johns Hopkins University, Baltimore, MD. MIM Number: 209900. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/

CHROMOSOMAL LOCATION

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

SOURCE

BBS3 (C-5) is a mouse monoclonal antibody raised against amino acids 67-186 mapping at the C-terminus of BBS3 of human origin.

PRODUCT

Each vial contains 200 µg IgG_{2a} kappa light chain 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-390021 X, 200 µg/0.1 ml.

BBS3 (C-5) is available conjugated to agarose (sc-390021 AC), 500 µg/ 0.25 ml agarose in 1 ml, for IP; to HRP (sc-390021 HRP), 200 µg/ml, for WB, IHC(P) and ELISA; to either phycoerythrin (sc-390021 PE), fluorescein (sc-390021 FITC), Alexa Fluor® 488 (sc-390021 AF488), Alexa Fluor® 546 (sc-390021 AF546), Alexa Fluor® 594 (sc-390021 AF594) or Alexa Fluor® 647 (sc-390021 AF647), 200 µg/ml, for WB (RGB), IF, IHC(P) and FCM; and to either Alexa Fluor® 680 (sc-390021 AF680) or Alexa Fluor® 790 (sc-390021 AF790), 200 µg/ml, for Near-Infrared (NIR) WB, IF and FCM.

APPLICATIONS

BBS3 (C-5) is recommended for detection of BBS3 of mouse, rat and human origin by Western Blotting (starting dilution 1:100, 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-5) is also recommended for detection of BBS3 in additional species, including canine and porcine.

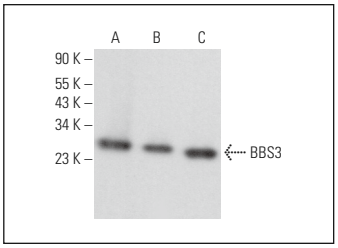
Suitable for use as control antibody for BBS3 siRNA (h): sc-60253, BBS3 siRNA (m): sc-60254, BBS3 shRNA Plasmid (h): sc-60253-SH, BBS3 shRNA Plasmid (m): sc-60254-SH, BBS3 shRNA (h) Lentiviral Particles: sc-60253-V and BBS3 shRNA (m) Lentiviral Particles: sc-60254-V.

BBS3 (C-5) X TransCruz antibody is recommended for Gel Supershift and ChIP applications.

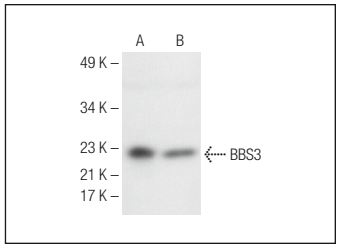
Molecular Weight of BBS3: 21 kDa.

Positive Controls: Hep G2 cell lysate: sc-2227, SH-SY5Y cell lysate: sc-3812 or WI-38 whole cell lysate: sc-364260.

DATA



BBS3 (C-5): sc-390021. Western blot analysis of BBS3 expression in SH-SY5Y (**A**), Y79 (**B**) and RPE-J (**C**) whole cell lysates.



BBS3 (C-5): sc-390021. Western blot analysis of BBS3 expression in WI-38 (**A**) and Hep G2 (**B**) whole cell lysates.

SELECT PRODUCT CITATIONS

1. Barbelanne, M., et al. 2015. Nephrocystin proteins NPHP5 and Cep290 regulate BBSome integrity, ciliary trafficking and cargo delivery. Hum. Mol. Genet. 24: 2185-2200.
2. Odabasi, E., et al. 2023. CCDC66 regulates primary cilium length and signaling via interactions with transition zone and axonemal proteins. J. Cell Sci. 136: jcs260327.

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.