## SANTA CRUZ BIOTECHNOLOGY, INC.

# BBS1 (F-1): sc-365138



#### 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 gene is ubiquitously expressed, with highest abundance in 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.
- Dollfus, H., et al. 2005. Update on Bardet-Biedl syndrome. J. Fr. Ophtalmol. 28: 106-112.
- 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.
- Hartmann, T.B., et al. 2005. SEREX identification of new tumor antigens linked to melanoma-associated retinopathy. Int. J. Cancer 114: 88-93.

### **CHROMOSOMAL LOCATION**

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

#### SOURCE

BBS1 (F-1) is a mouse monoclonal antibody raised against amino acids 291-590 mapping near the C-terminus of BBS1 of human origin.

#### PRODUCT

Each vial contains 200  $\mu$ g lgG<sub>1</sub> 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-365138 X, 200  $\mu$ g/0.1 ml.

BBS1 (F-1) is available conjugated to agarose (sc-365138 AC), 500 µg/0.25 ml agarose in 1 ml, for IP; to HRP (sc-365138 HRP), 200 µg/ml, for WB, IHC(P) and ELISA; to either phycoerythrin (sc-365138 PE), fluorescein (sc-365138 FITC), Alexa Fluor<sup>®</sup> 488 (sc-365138 AF488), Alexa Fluor<sup>®</sup> 546 (sc-365138 AF546), Alexa Fluor<sup>®</sup> 594 (sc-365138 AF594) or Alexa Fluor<sup>®</sup> 647 (sc-365138 AF647), 200 µg/ml, for WB (RGB), IF, IHC(P) and FCM; and to either Alexa Fluor<sup>®</sup> 680 (sc-365138 AF680) or Alexa Fluor<sup>®</sup> 790 (sc-365138 AF790), 200 µg/ml, for Near-Infrared (NIR) WB, IF and FCM.

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#### APPLICATIONS

BBS1 (F-1) is recommended for detection of BBS1 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), 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 (F-1) is also recommended for detection of BBS1 in additional species, including equine, canine, bovine and porcine.

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 (F-1) X TransCruz antibody is recommended for Gel Supershift and ChIP applications.

Molecular Weight of BBS1: 65 kDa.

Positive Controls: mouse brain extract: sc-2253.

#### DATA





BBS1 (F-1): sc-365138. Western blot analysis of BBS1 expression in mouse brain tissue extract.

BBS1 (F-1): sc-365138. Immunoperoxidase staining of formalin fixed, parafin-embedded human lymph node tissue showing cytoplasmic staining of cells in germinal and non-germinal centers.

## SELECT PRODUCT CITATIONS

 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, \*\*D0 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.

## PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.