SANTA CRUZ BIOTECHNOLOGY, INC.

BBS8 (Q30): sc-100682

BACKGROUND

Bardet-Biedl syndrome (BBS) is a heterogeneous pleiotropic genetic disorder characterized by obesity, photoreceptor degeneration, polydactyly, hypogenitalism, renal abnormalities, developmental delay, diabetes, hypertension and congenital heart defects. 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. Mutations in BBS8, also designated tetratricopeptide repeat protein (TTC8), probably account for only a minority (2%) of BBS families, underlining the difficulty of genotyping heterogeneous conditions. The identification of BBS8 provides the key to the pathogenesis of the condition as a primary ciliary disorder.

REFERENCES

1. Heon, E., Westall, C., Carmi, R., Elbedour, K., Panton, C., Mackeen, L., Stone, E.M. and Sheffield, V.C. 2005. Ocular phenotypes of three genetic variants of Bardet-Biedl syndrome. Am. J. Med. Genet. A. 132: 283-287.
2. Nakane, T. and Biesecker L.G. 2005. No evidence for triallelic inheritance of MKKS/BBS loci in Amish Mckusick-Kaufman syndrome. Am. J. Med. Genet. A. 138: 32-34.
3. Hichri, H., Stoetzel, C., Laurier, V., Caron, S., Sigaudy, S., Sarda, P., Hamel, C., Martin-Coignard, D., Gilles, M., Leheup, B., Holder, M., Kaplan, J., Bitoun, P., Lacombe, D., Verloes, A., Bonneau, D., Perrin-Schmitt, F., Brandt, C., Besancon, A.F., Mandel, J.L., Cossee, M. and Dollfus, H. 2005. Testing for triallelism: analysis of six BBS genes in a Bardet-Biedl syndrome family cohort. Eur. J. Hum. Genet. 13: 607-616.
4. Dollfus, H., Verloes, A., Bonneau, D., Cossee, M., Perrin-Schmitt, F., Brandt, C., Flament, J. and Mandel, J.L. 2005. Update on Bardet-Biedl syndrome. J. Fr. Ophtalmol. 28: 106-112.

CHROMOSOMAL LOCATION

Genetic locus: TTC8 (human) mapping to 14q31.3; Ttc8 (mouse) mapping to 12 E.

SOURCE

BBS8 (Q30) is a mouse monoclonal antibody raised against recombinant BBS8 of human origin.

PRODUCT

Each vial contains 100 µg IgG_{2a} in 1.0 ml PBS with < 0.1% sodium azide and 0.1% gelatin.

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 or our catalog for detailed protocols and support products.

APPLICATIONS

BBS8 (Q30) is recommended for detection of BBS8 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)] and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for BBS8 siRNA (h): sc-60261, BBS8 siRNA (m): sc-60262, BBS8 shRNA Plasmid (h): sc-60261-SH, BBS8 shRNA Plasmid (m): sc-60262-SH, BBS8 shRNA (h) Lentiviral Particles: sc-60261-V and BBS8 shRNA (m) Lentiviral Particles: sc-60262-V.

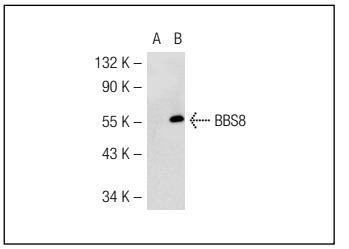
Molecular Weight of BBS8: 60 kDa.

Positive Controls: NIH/3T3 whole cell lysate: sc-2210.

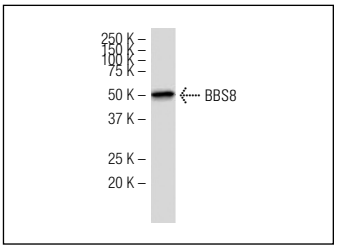
RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-mouse IgG-HRP: sc-2005 (dilution range: 1:2000-1:32,000) or Cruz Marker™ compatible goat anti-mouse IgG-HRP: sc-2031 (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).

DATA



BBS8 (Q30): sc-100682. Western blot analysis of BBS8 expression in non-transfected: sc-117752 (**A**) and mouse BBS8 transfected: sc-118690 (**B**) 293T whole cell lysates.



BBS8 (Q30): sc-100682. Western blot analysis of BBS8 expression in NIH/3T3 whole cell lysate.

RESEARCH USE

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