

## BBS8 (H-300): sc-99095

### BACKGROUND

Bardet-Biedl syndrome (BBS) is a heterogeneous pleiotropic genetic disorder characterized by obesity, photoreceptor degeneration, polydactyly, hypogonitalism, 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. Young, J.C., Obermann, W.M. and Hartl, F.U. 1998. Specific binding of tetratricopeptide repeat proteins to the C-terminal 12-kDa domain of HSP 90. *J. Biol. Chem.* 273: 18007-18010.
2. Cortajarena, A.L., Kajander, T., Pan, W., Cocco, M.J. and Regan, L. 2004. Protein design to understand peptide ligand recognition by tetratricopeptide repeat proteins. *Protein Eng. Des. Sel.* 17: 399-409.
3. Heon, E., Westall, C., Carmi, R., Elbedour, K., Pantan, 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.
4. 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.
5. 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.
6. 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.
7. Nachury, M.V., Loktev, A.V., Zhang, Q., Westlake, C.J., Peränen, J., Merdes, A., Slusarski, D.C., Scheller, R.H., Bazan, J.F., Sheffield, V.C. and Jackson, P.K. 2007. A core complex of BBS proteins cooperates with the GTPase Rab 8 to promote ciliary membrane biogenesis. *Cell* 129: 1201-1213.

### CHROMOSOMAL LOCATION

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

### SOURCE

BBS8 (H-300) is a rabbit polyclonal antibody raised against amino acids 232-531 mapping at the C-terminus of BBS8 of human origin.

### RESEARCH USE

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

### 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-99095 X, 200 µg/0.1 ml.

### APPLICATIONS

BBS8 (H-300) 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)], 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).

BBS8 (H-300) is also recommended for detection of BBS8 in additional species, including equine, canine, bovine, porcine and avian.

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.

BBS8 (H-300) X TransCruz antibody is recommended for Gel Supershift and ChIP applications.

Molecular Weight of BBS8: 62 kDa.

Positive Controls: NIH/3T3 whole cell lysate: sc-2210, LNCaP cell lysate: sc-2231 or mouse liver extract: sc-2256.

### RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (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 goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

### STORAGE

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


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Try **BBS8 (E-2): sc-271009**, our highly recommended monoclonal alternative to BBS8 (H-300).