

BBS4 (H-304): sc-67201

BACKGROUND

Bardet-Biedl syndrome (BBS) is a pleiotropic genetic disorder characterized by obesity, photoreceptor degeneration, polydactyly, hypogonadism, renal abnormalities and developmental delay. Other associated clinical findings in BBS patients include 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. BBS4 is expressed in the olfactory epithelium and localizes to the centriolar satellites of centrosomes and basal bodies of primary cilia. BBS4 regulates the p150 subunit of the Dynein transport machinery (DCTN1) to attract pericentriolar material-1 protein (PCM1) and its associated components to the satellites. Loss of BBS4 is correlated with obesity caused by abnormal lipid profiles, liver dysfunction, elevated Insulin and abnormal leptin levels.

REFERENCES

- Hichri, H., et al. 2005. Testing for triallelism: analysis of six BBS genes in a Bardet-Biedl syndrome family cohort. *Eur. J. Hum. Genet.* 13: 607-616.
- Lee, S., et al. 2005. Essential role for the Prader-Willi syndrome protein necln in axonal outgrowth. *Hum. Mol. Genet.* 14: 627-637.
- Dollfus, H., et al. 2005. Update on Bardet-Biedl syndrome. *J. Fr. Ophthalmol.* 28: 106-112.
- Karmous-Benailly, H., et al. 2005. Antenatal presentation of Bardet-Biedl syndrome may mimic Meckel syndrome. *Am. J. Hum. Genet.* 76: 493-504.
- Ahmad, J., et al. 2005. DFN48, a new nonsyndromic recessive deafness locus, maps to chromosome 15q23-q25.1. *Hum. Genet.* 116: 407-412.
- Heon, E., et al. 2005. Ocular phenotypes of three genetic variants of Bardet-Biedl syndrome. *Am. J. Med. Genet. A* 132: 283-287.
- Iannaccone, A., et al. 2005. Clinical evidence of decreased olfaction in Bardet-Biedl syndrome caused by a deletion in the BBS4 gene. *Am. J. Med. Genet. A* 132: 343-346.

CHROMOSOMAL LOCATION

Genetic locus: BBS4 (human) mapping to 15q24.1; Bbs4 (mouse) mapping to 9 B.

SOURCE

BBS4 (H-304) is a rabbit polyclonal antibody raised against amino acids 33-336 mapping near the N-terminus of BBS4 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.

STORAGE

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

APPLICATIONS

BBS4 (H-304) is recommended for detection of BBS4 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), 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).

BBS4 (H-304) is also recommended for detection of BBS4 in additional species, including equine, canine, bovine and porcine.

Suitable for use as control antibody for BBS4 siRNA (h): sc-60255, BBS4 siRNA (m): sc-60256, BBS4 shRNA Plasmid (h): sc-60255-SH, BBS4 shRNA Plasmid (m): sc-60256-SH, BBS4 shRNA (h) Lentiviral Particles: sc-60255-V and BBS4 shRNA (m) Lentiviral Particles: sc-60256-V.

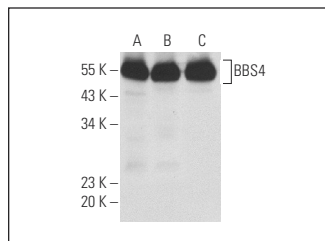
Molecular Weight of BBS4: 58 kDa.

Positive Controls: mouse heart extract: sc-2254, rat heart extract: sc-2393 or rat kidney extract: sc-2394.

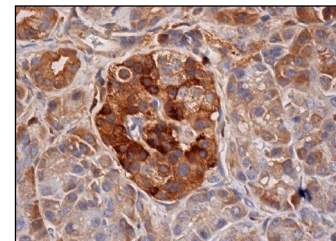
RECOMMENDED SECONDARY REAGENTS

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. 4) Immunohistochemistry: use ImmunoCruz™: sc-2051 or ABC: sc-2018 rabbit IgG Staining Systems.

DATA



BBS4 (H-304): sc-67201. Western blot analysis of BBS4 expression in mouse heart (A), rat heart (B) and rat kidney (C) tissue extracts.



BBS4 (H-304): sc-67201. Immunoperoxidase staining of formalin fixed, paraffin-embedded human pancreas tissue showing cytoplasmic staining of exocrine glandular cells and Islets of Langerhans.

RESEARCH USE

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