



BBS1 siRNA (h): sc-60249

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 is the protein most commonly involved in Bardet-Biedl syndrome. The BBS1 gene is ubiquitously expressed, with highest abundance 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. Myktyyn, K., et al. 2003. Evaluation of complex locus (BBS1). *Am. J. Hum. Genet.* 72: 429-437.
3. Dollfus, H., et al. 2005. Update on Bardet-Biedl syndrome. *J. Fr. Ophthalmol.* 28: 106-112.
4. 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.
5. Hartmann, T.B., et al. 2005. SEREX identification of new tumor antigens linked to melanoma-associated retinopathy. *Int. J. Cancer* 114: 88-93.
6. Heon, E., et al. 2005. Ocular phenotypes of three genetic variants of Bardet-Biedl syndrome. *Am. J. Med. Genet. A* 132: 283-287.
7. 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.

CHROMOSOMAL LOCATION

Genetic locus: BBS1 (human) mapping to 11q13.2.

PRODUCT

BBS1 siRNA (h) is a pool of 3 target-specific 19-25 nt siRNAs designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10 μ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see BBS1 shRNA Plasmid (h): sc-60249-SH and BBS1 shRNA (h) Lentiviral Particles: sc-60249-V as alternate gene silencing products.

For independent verification of BBS1 (h) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-60249A, sc-60249B and sc-60249C.

PROTOCOLS

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

STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20° C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20° C, avoid contact with RNases and repeated freeze thaw cycles.

Resuspend lyophilized siRNA duplex in 330 μ l of the RNase-free water provided. Resuspension of the siRNA duplex in 330 μ l of RNase-free water makes a 10 μ M solution in a 10 μ M Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

APPLICATIONS

BBS1 siRNA (h) is recommended for the inhibition of BBS1 expression in human cells.

SUPPORT REAGENTS

For optimal siRNA transfection efficiency, Santa Cruz Biotechnology's siRNA Transfection Reagent: sc-29528 (0.3 ml), siRNA Transfection Medium: sc-36868 (20 ml) and siRNA Dilution Buffer: sc-29527 (1.5 ml) are recommended. Control siRNAs or Fluorescein Conjugated Control siRNAs are available as 10 μ M in 66 μ l. Each contain a scrambled sequence that will not lead to the specific degradation of any known cellular mRNA. Fluorescein Conjugated Control siRNAs include: sc-36869, sc-44239, sc-44240 and sc-44241. Control siRNAs include: sc-37007, sc-44230, sc-44231, sc-44232, sc-44233, sc-44234, sc-44235, sc-44236, sc-44237 and sc-44238.

GENE EXPRESSION MONITORING

BBS1 (F-1): sc-365138 is recommended as a control antibody for monitoring of BBS1 gene expression knockdown by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000) or immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500).

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgG κ BP-HRP: sc-516102 or m-IgG κ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use m-IgG κ BP-FITC: sc-516140 or m-IgG κ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor BBS1 gene expression knockdown using RT-PCR Primer: BBS1 (h)-PR: sc-60249-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

RESEARCH USE

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