

BBS3 siRNA (h): sc-60253

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; BBS genes map to eight genetic loci and encode eight proteins, BBS1-8. Five BBS genes encode basal body or cilia proteins, suggesting that BBS is a ciliary dysfunction disorder. Bardet-Biedl syndrome-3 (BBS3) results from a homozygous C-to-T transition in exon 7 of the ARL6 gene, resulting in an Arg 122-to-Ter mutation with a premature truncation of the protein from 186 to 121 amino acids. Heterozygosity in a mutation of the BBS3 gene modifies the expression of the Met 390-to-Arg mutation in the BBS1 gene.

REFERENCES

1. Bruford, E.A., et al. 1997. Linkage mapping in 29 Bardet-Biedl syndrome families confirms loci in chromosomal regions 11q13, 15q22.3-q23, and 16q21. *Genomics* 411: 93-99.
2. Young, T.L., et al. 1998. Canadian Bardet-Biedl syndrome family reduces the critical region of BBS3 (3p) and presents with a variable phenotype. *Am. J. Med. Genet.* 78: 461-467.
3. Ghadami, M., et al. 2000. Bardet-Biedl syndrome type 3 in an Iranian family: clinical study and confirmation of disease localization. *Am. J. Med. Genet.* 94: 433-437.
4. Heon, E., et al. 2005. Ocular phenotypes of three genetic variants of Bardet-Biedl syndrome. *Am. J. Med. Genet. A* 132A: 283-287.
5. 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.
6. Online Mendelian Inheritance in Man, OMIM[™]. 2008. Johns Hopkins University, Baltimore, MD. MIM Number: 209900. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>

CHROMOSOMAL LOCATION

Genetic locus: ARL6 (human) mapping to 3q11.2.

PRODUCT

BBS3 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 BBS3 shRNA Plasmid (h): sc-60253-SH and BBS3 shRNA (h) Lentiviral Particles: sc-60253-V as alternate gene silencing products.

For independent verification of BBS3 (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-60253A, sc-60253B and sc-60253C.

RESEARCH USE

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

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

BBS3 siRNA (h) is recommended for the inhibition of BBS3 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

BBS3 (C-5): sc-390021 is recommended as a control antibody for monitoring of BBS3 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 BBS3 gene expression knockdown using RT-PCR Primer: BBS3 (h)-PR: sc-60253-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

PROTOCOLS

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