

# PHF6 siRNA (h): sc-90882

## BACKGROUND

Zinc-finger proteins contain DNA-binding domains and have a wide variety of functions, most of which encompass some form of transcriptional activation or repression. PHF6 (PHD finger protein 6), also known as BORJ, is a 365 amino acid protein that localizes to the nucleus and contains two PHD-type zinc fingers. Expressed ubiquitously, PHF6 exists as two alternatively spliced isoforms and is thought to play a role in transcriptional regulation. Upon DNA damage, PHF6 is subject to phosphorylation, probably by ATM or ATR. Mutations in the gene encoding PHF6 are the cause of Börjeson-Forssman-Lehmann syndrome (BFLS), an X-linked recessive disorder that is characterized by mental retardation, epilepsy, hypogonadism, hypometabolism, obesity with marked gynecomastia, swelling of subcutaneous tissue of the face and narrow palpebral fissure.

## REFERENCES

1. Börjeson, M., et al. 1962. An X-linked, recessively inherited syndrome characterized by grave mental deficiency, epilepsy, and endocrine disorder. *Acta Med. Scand.* 171: 13-21.
2. Lower, K.M., et al. 2002. Mutations in PHF6 are associated with Börjeson-Forssman-Lehmann syndrome. *Nat. Genet.* 32: 661-665.
3. Lower, K.M., et al. 2004. 1024C> T (R342X) is a recurrent PHF6 mutation also found in the original Börjeson-Forssman-Lehmann syndrome family. *Eur. J. Hum. Genet.* 12: 787-789.
4. Vallee, D., et al. 2004. A novel PHF6 mutation results in enhanced exon skipping and mild Börjeson-Forssman-Lehmann syndrome. *J. Med. Genet.* 41: 778-783.
5. Crawford, J., et al. 2006. Mutation screening in Börjeson-Forssman-Lehmann syndrome: identification of a novel *de novo* PHF6 mutation in a female patient. *J. Med. Genet.* 43: 238-243.
6. Voss, A.K., et al. 2007. Protein and gene expression analysis of Phf6, the gene mutated in the Börjeson-Forssman-Lehmann syndrome of intellectual disability and obesity. *Gene Expr. Patterns* 7: 858-871.

## CHROMOSOMAL LOCATION

Genetic locus: PHF6 (human) mapping to Xq26.2.

## PRODUCT

PHF6 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  $\mu$ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see PHF6 shRNA Plasmid (h): sc-90882-SH and PHF6 shRNA (h) Lentiviral Particles: sc-90882-V as alternate gene silencing products.

For independent verification of PHF6 (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-90882A, sc-90882B and sc-90882C.

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

## APPLICATIONS

PHF6 siRNA (h) is recommended for the inhibition of PHF6 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  $\mu$ M in 66  $\mu$ 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

PHF6 (H-4): sc-365237 is recommended as a control antibody for monitoring of PHF6 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 $\kappa$  BP-HRP: sc-516102 or m-IgG $\kappa$  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 $\kappa$  BP-FITC: sc-516140 or m-IgG $\kappa$  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 PHF6 gene expression knockdown using RT-PCR Primer: PHF6 (h)-PR: sc-90882-PR (20  $\mu$ 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](http://www.scbt.com) for detailed protocols and support products.