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# SMCHD1 siRNA (h): sc-156012

## BACKGROUND

SMCHD1 (structural maintenance of chromosomes flexible hinge domain containing 1) is a 2,005 amino acid protein and novel modifier of epigenetic gene silencing. SMCHD1 may have a role in the hypermethylation of CpG islands associated with the maintenance of X chromosome inactivation. SMCHD1 exists as three alternatively spliced isoforms and is encoded by a gene located on human chromosome 18p11.32, which houses over 300 protein-coding genes and contains nearly 76 million bases, representing about 2.5% of total DNA in cells. There are a variety of diseases associated with defects in chromosome 18-localized genes, some of which include Niemann-Pick disease, hereditary hemorrhagic telangiectasia, erythropoietic protoporphyria and follicular lymphomas. Also, three chromosomal abnormalities result from meiotic nondisjunction events of chromosome 18: Monosomy 18p, Trisomy 18 (also known as Edwards syndrome) and Tetrasomy 18p. The LOC390856 gene product has been provisionally designated LOC390856 pending further characterization.

## REFERENCES

- Grosso, S., Pucci, L., Di Bartolo, R.M., Gobbi, G., Bartalini, G., Anichini, C., Scarinci, R., Balestri, M., Farnetani, M.A., Cioni, M., Morgese, G. and Balestri, P. 2005. Chromosome 18 aberrations and epilepsy: a review. *Am. J. Med. Genet. A* 134A: 88-94.
- Cerminara, C., Lo Castro, A., D'Argenzio, L., Galasso, C. and Curatolo, P. 2008. Epilepsy and deletion syndromes of chromosome 18: do not forget the short arm! *Epilepsia* 49: 1813-1814.
- Ashe, A., Morgan, D.K., Whitelaw, N.C., Bruxner, T.J., Vickaryous, N.K., Cox, L.L., Butterfield, N.C., Wicking, C., Blewitt, M.E., Wilkins, S.J., Anderson, G.J., Cox, T.C. and Whitelaw, E. 2008. A genome-wide screen for modifiers of transgene variegation identifies genes with critical roles in development. *Genome Biol.* 9: R182.
- Kohan, R. and Bower, C. 2008. Improving the health care experiences of families given the prenatal diagnosis of Trisomy 18. *J. Perinatol.* 28: 719.
- Blewitt, M.E., Gendrel, A.V., Pang, Z., Sparrow, D.B., Whitelaw, N., Craig, J.M., Apedaile, A., Hilton, D.J., Dunwoodie, S.L., Brockdorff, N., Kay, G.F. and Whitelaw, E. 2008. SMCHD1, containing a structural-maintenance-of-chromosomes hinge domain, has a critical role in X inactivation. *Nat. Genet.* 40: 663-669.
- Shaw, J. 2008. Trisomy 18: a case study. *Neonatal Netw.* 27: 33-41.
- Turleau, C. 2008. Monosomy 18p. *Orphanet. J. Rare Dis.* 3: 4.
- Witters, I. and Fryns, J.P. 2008. Trisomy 18 presenting with severe limb deformations. *Prenat. Diagn.* 28: 549-550.
- Edwards, S. and Waters, J.J. 2008. Prenatal diagnosis of monosomy 18p involving a jumping translocation. *Prenat. Diagn.* 28: 764-766.

## CHROMOSOMAL LOCATION

Genetic locus: SMCHD1 (human) mapping to 18p11.32.

## RESEARCH USE

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

## PRODUCT

SMCHD1 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 SMCHD1 shRNA Plasmid (h): sc-156012-SH and SMCHD1 shRNA (h) Lentiviral Particles: sc-156012-V as alternate gene silencing products.

For independent verification of SMCHD1 (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-156012A, sc-156012B and sc-156012C.

## 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

SMCHD1 siRNA (h) is recommended for the inhibition of SMCHD1 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.

## RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor SMCHD1 gene expression knockdown using RT-PCR Primer: SMCHD1 (h)-PR: sc-156012-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.