

TMEM194 siRNA (h): sc-95900

BACKGROUND

TMEM194 (transmembrane protein 194A) is a 444 amino acid multi-pass membrane protein that is expressed as two alternatively spliced isoforms. The gene encoding TMEM194 maps to human chromosome 12, which encodes over 1,100 genes within 132 million bases and makes up about 4.5% of the human genome. A number of skeletal deformities are linked to chromosome 12 including hypochondrogenesis, achondrogenesis and Kniest dysplasia. Noonan syndrome, which includes heart and facial developmental defects among the primary symptoms, is caused by a mutant form of PTPN11 gene product, SH-PTP2. Chromosome 12 is also home to a homeobox gene cluster which encodes crucial transcription factors for morphogenesis, and the natural killer complex gene cluster encoding C-type lectin proteins which mediate the NK cell response to MHC I interaction. Trisomy 12p leads to facial development defects, seizure disorders and a host of other symptoms varying in severity depending on the extent of mosaicism and is most severe in cases of complete trisomy.

REFERENCES

1. Allen, T.L., Brothman, A.R., Carey, J.C. and Chance, P.F. 1996. Cytogenetic and molecular analysis in trisomy 12p. *Am. J. Med. Genet.* 63: 250-256.
2. Yang, W. and Cole, W.G. 1998. Low basal transcripts of the COL2A1 collagen gene from lymphoblasts show alternative splicing of exon 12 in the Kniest form of spondyloepiphyseal dysplasia. *Hum. Mutat. Suppl.* 1: S1-S2.
3. Trowsdale, J., Barten, R., Haude, A., Stewart, C.A., Beck, S. and Wilson, M.J. 2001. The genomic context of natural killer receptor extended gene families. *Immunol. Rev.* 181: 20-38.
4. Zumkeller, W., Volleth, M., Muschke, P., Tönnies, H., Heller, A., Liehr, T., Wieacker, P. and Stumm, M. 2004. Genotype/phenotype analysis in a patient with pure and complete trisomy 12p. *Am. J. Med. Genet. A* 129A: 261-264.
5. Nishimura, G., Haga, N., Kitoh, H., Tanaka, Y., Sonoda, T., Kitamura, M., Shirahama, S., Itoh, T., Nakashima, E., Ohashi, H. and Ikegawa, S. 2005. The phenotypic spectrum of COL2A1 mutations. *Hum. Mutat.* 26: 36-43.
6. Kelley, J., Walter, L. and Trowsdale, J. 2005. Comparative genomics of natural killer cell receptor gene clusters. *PLoS Genet.* 1: 129-139.
7. Segel, R., Peter, I., Demmer, L.A., Cowan, J.M., Hoffman, J.D. and Bianchi, D.W. 2006. The natural history of trisomy 12p. *Am. J. Med. Genet. A* 140: 695-703.
8. Shchelochkov, O.A., Patel, A., Weissenberger, G.M., Chinault, A.C., Wiszniewska, J., Fernandes, P.H., Eng, C., Kukulich, M.K. and Sutton, V.R. 2008. Duplication of chromosome band 12q24.11q24.23 results in apparent Noonan syndrome. *Am. J. Med. Genet. A* 146A: 1042-1048.

CHROMOSOMAL LOCATION

Genetic locus: NEMP1 (human) mapping to 12q13.3.

PROTOCOLS

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

PRODUCT

TMEM194 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 TMEM194 shRNA Plasmid (h): sc-95900-SH and TMEM194 shRNA (h) Lentiviral Particles: sc-95900-V as alternate gene silencing products.

For independent verification of TMEM194 (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-95900A, sc-95900B and sc-95900C.

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

TMEM194 siRNA (h) is recommended for the inhibition of TMEM194 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.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor TMEM194 gene expression knockdown using RT-PCR Primer: TMEM194 (h)-PR: sc-95900-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.