

# NCKAP5L siRNA (h): sc-95803

## BACKGROUND

NCKAP5L (NCK-associated protein 5-like) is a 1,330 amino acid protein that exists as four alternatively spliced isoforms. The gene encoding NCKAP5L maps to human chromosome 12, which encodes over 1,100 genes within 132 million bases, and comprises 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 which vary in severity depending on the extent of mosaicism. It 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. Delgado Carrasco, J., Casanova Morcillo, A., Zabalza Alvillos, M. and Ayala Garcés, A. 2001. Achondrogenesis type II-hypochondrogenesis: radiological features. *Case report. An. Esp. Pediatr.* 55: 553-557.
3. Yokoyama, T., Nakatani, S. and Murakami, A. 2003. A case of Kniest dysplasia with retinal detachment and the mutation analysis. *Am. J. Ophthalmol.* 136: 1186-1188.
4. Forzano, F., Lituania, M., Viassolo, A., Superti-Furga, V., Wildhardt, G., Zabel, B. and Faravelli, F. 2007. A familial case of achondrogenesis type II caused by a dominant COL2A1 mutation and "patchy" expression in the mosaic father. *Am. J. Med. Genet. A* 143A: 2815-2820.
5. Wainwright, H. and Beighton, P. 2008. Visceral manifestations of hypochondrogenesis. *Virchows Arch.* 453: 203-207.
6. Lo, F.S., Luo, J.D., Lee, Y.J., Shu, S.G., Kuo, M.T. and Chiou, C.C. 2009. High resolution melting analysis for mutation detection for PTPN11 gene: applications of this method for diagnosis of Noonan syndrome. *Clin. Chim. Acta* 409: 75-77.
7. Benussi, D.G., Costa, P., Zollino, M., Murdolo, M., Petix, V., Carrozzi, M. and Pecile, V. 2009. Trisomy 12p and monosomy 4p: phenotype-genotype correlation. *Genet. Test. Mol. Biomarkers* 13: 199-204.

## CHROMOSOMAL LOCATION

Genetic locus: NCKAP5L (human) mapping to 12q13.12.

## PRODUCT

NCKAP5L siRNA (h) is a target-specific 19-25 nt siRNA 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 NCKAP5L shRNA Plasmid (h): sc-95803-SH and NCKAP5L shRNA (h) Lentiviral Particles: sc-95803-V as alternate gene silencing 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  $\mu$ 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

NCKAP5L siRNA (h) is recommended for the inhibition of NCKAP5L 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 NCKAP5L gene expression knockdown using RT-PCR Primer: NCKAP5L (h)-PR: sc-95803-PR (20  $\mu$ 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.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) for detailed protocols and support products.