

NCKAP5L (N-15): sc-248057

BACKGROUND

NCKAP5L (NCK-associated protein 5-like) is a 1,330 amino acid protein that exists as 4 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

- 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.
- Delgado Carrasco, J., Casanova Morcillo, A., Zabalza Alvillos, M. and Ayala Garces, A. 2001. Achondrogenesis type II-hypochondrogenesis: radiological features. *Case report. An. Esp. Pediatr.* 55: 553-557.
- 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.
- 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.
- Wainwright, H. and Beighton, P. 2008. Visceral manifestations of hypochondrogenesis. *Virchows Arch.* 453: 203-207.
- 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.
- 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.

SOURCE

NCKAP5L (N-15) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the N-terminus of NCKAP5L of human origin.

STORAGE

Store at 4° C, ****DO NOT FREEZE****. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-248057 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

NCKAP5L (N-15) is recommended for detection of NCKAP5L of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for NCKAP5L siRNA (h): sc-95803, NCKAP5L shRNA Plasmid (h): sc-95803-SH and NCKAP5L shRNA (h) Lentiviral Particles: sc-95803-V.

Molecular Weight of NCKAP5L isoforms: 139/135/11/137 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

RESEARCH USE

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

PROTOCOLS

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