# SANTA CRUZ BIOTECHNOLOGY, INC.

# CCDC92 (N-18): sc-242380



## BACKGROUND

CCDC92 (coiled-coil domain-containing protein 92) is a 331 amino acid protein encoded by a gene that maps to human chromosome 12q24.31. Encoding over 1,100 genes within 132 million bases, chromosome 12 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 that mediate the NK cell response to MHC I interaction. Trisomy 12p leads to facial development defects, seizure disorders and a host of other symptoms that vary in severity depending on the extent of mosaicism. It is most severe in cases of complete trisomy.

## REFERENCES

- 1. Allen, T.L., et al. 1996. Cytogenetic and molecular analysis in trisomy 12p. Am. J. Med. Genet. 63: 250-256.
- Delgado Carrasco, J., et al. 2001. Achondrogenesis type II-hypochondrogenesis: radiological features.Case report. An. Esp. Pediatr. 55: 553-557.
- Yokoyama, T., et al. 2003. A case of Kniest dysplasia with retinal detachment and the mutation analysis. Am. J. Ophthalmol. 136: 1186-1188.
- Forzano, F., et al. 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 143: 2815-2820.
- Wainwright, H. and Beighton, P. 2008. Visceral manifestations of hypochondrogenesis. Virchows Arch. 453: 203-207.
- Lo, F.S., et al. 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.

## CHROMOSOMAL LOCATION

Genetic locus: CCDC92 (human) mapping to 12q24.31.

## SOURCE

CCDC92 (N-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the N-terminus of CCDC92 of human origin.

## PRODUCT

Each vial contains 200  $\mu g$  lgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-242380 P, (100  $\mu$ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

#### STORAGE

Store at 4° C, \*\*D0 NOT FREEZE\*\*. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

### APPLICATIONS

CCDC92 (N-18) is recommended for detection of CCDC92 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); non cross-reactive with other CCDC family proteins.

Suitable for use as control antibody for CCDC92 siRNA (h): sc-95990, CCDC92 shRNA Plasmid (h): sc-95990-SH and CCDC92 shRNA (h) Lentiviral Particles: sc-95990-V.

Molecular Weight of CCDC92: 37 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<sup>™</sup> compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker<sup>™</sup> Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluo-rescence: 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<sup>™</sup> 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.