

# FAM130A1 (C-18): sc-242682

## BACKGROUND

FAM130A1, also designated Cysteine/serine-rich nuclear protein 2 (CSRNP-2) and TGF- $\beta$ -induced apoptosis protein 12 (TAIP12), is a 543 amino acid protein which is encoded by a gene that maps to human chromosome 12. 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 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

- Allen, T.L., et al. 1996. Cytogenetic and molecular analysis in trisomy 12p. *Am. J. Med. Genet.* 63: 250-256.
- 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.
- Trowsdale, J., et al. 2001. The genomic context of natural killer receptor extended gene families. *Immunol. Rev.* 181: 20-38.
- Lee, S.Y., et al. 2003. Immunomic analysis of human sarcoma. *Proc. Natl. Acad. Sci. USA* 100: 2651-2656.
- Zumkeller, W., et al. 2004. Genotype/phenotype analysis in a patient with pure and complete trisomy 12p. *Am. J. Med. Genet. A* 129: 261-264.
- Lee, L.A., et al. 2005. *Drosophila* genome-scale screen for PAN GU kinase substrates identifies Mat89Bb as a cell cycle regulator. *Dev. Cell* 8: 435-442.
- Kelley, J., et al. 2005. Comparative genomics of natural killer cell receptor gene clusters. *PLoS Genet.* 1: e27.
- Nishimura, G., et al. 2005. The phenotypic spectrum of COL2A1 mutations. *Hum. Mutat.* 26: 36-43.
- Segel, R., et al. 2006. The natural history of trisomy 12p. *Am. J. Med. Genet. A* 140: 695-703.

## CHROMOSOMAL LOCATION

Genetic locus: CSRNP2 (human) mapping to 12q13.12; Csrnp2 (mouse) mapping to 15 F1.

## SOURCE

FAM130A1 (C-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the C-terminus of FAM130A1 of human origin.

## PRODUCT

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

Blocking peptide available for competition studies, sc-242682 P, (100  $\mu$ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

FAM130A1 (C-18) is recommended for detection of FAM130A1 of mouse and 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 FAM130A2.

Suitable for use as control antibody for FAM130A1 siRNA (h): sc-96103, FAM130A1 siRNA (m): sc-145026, FAM130A1 shRNA Plasmid (h): sc-96103-SH, FAM130A1 shRNA Plasmid (m): sc-145026-SH, FAM130A1 shRNA (h) Lentiviral Particles: sc-96103-V and FAM130A1 shRNA (m) Lentiviral Particles: sc-145026-V.

Molecular Weight of FAM130A1: 60 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.

## STORAGE

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

## 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) or our catalog for detailed protocols and support products.