

# C12orf53 (C-20): sc-242026

## BACKGROUND

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 class 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. The C12orf53 gene product has been provisionally designated C12orf53 pending further characterization.

## REFERENCES

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- Trowsdale, J., et al. 2001. The genomic context of natural killer receptor extended gene families. *Immunol. Rev.* 181: 20-38.
- Zumkeller, W., et al. 2004. Genotype/phenotype analysis in a patient with pure and complete trisomy 12p. *Am. J. Med. Genet. A* 129A: 261-264.
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- 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* 140A: 695-703.
- Stein, R. 2007. Genetics of Noonan syndrome—a new gene, and the search is still on. *Clin. Genet.* 72: 402-404.

## CHROMOSOMAL LOCATION

Genetic locus: PIANP (human) mapping to 12p13.31; Pianp (mouse) mapping to 6 F2.

## SOURCE

C12orf53 (C-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping within a C-terminal cytoplasmic domain of C12orf53 of human origin.

## 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-242026 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

C12orf53 (C-20) is recommended for detection of C12orf53 of human origin, C530028021Rik of mouse origin and the corresponding rat homolog by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], 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).

C12orf53 (C-20) is also recommended for detection of C12orf53 in additional species, including equine, bovine and porcine.

Suitable for use as control antibody for C12orf53 siRNA (h): sc-96043, C530028021Rik siRNA (m): sc-141916, C12orf53 shRNA Plasmid (h): sc-96043-SH, C530028021Rik shRNA Plasmid (m): sc-141916-SH, C12orf53 shRNA (h) Lentiviral Particles: sc-96043-V and C530028021Rik shRNA (m) Lentiviral Particles: sc-141916-V.

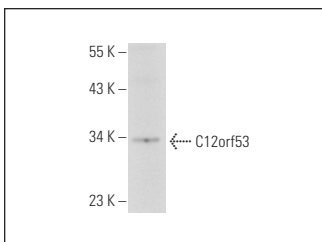
Molecular Weight of C12orf53: 30 kDa.

Positive Controls: mouse liver extract: sc-2256 or c4 whole cell lysate: sc-364186.

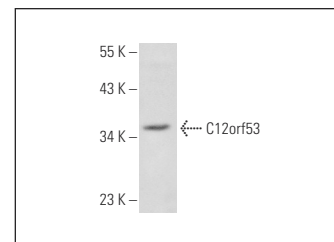
## RECOMMENDED SECONDARY REAGENTS

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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) 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.

## DATA



C12orf53 (C-20): sc-242026. Western blot analysis of C12orf53 expression in mouse liver tissue extract.



C12orf53 (C-20): sc-242026. Western blot analysis of C12orf53 expression in c4 whole cell lysate.

## 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.