

# C12orf48 (Y-18): sc-245199

## 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 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 C12orf48 gene product has been provisionally designated C12orf48 pending further characterization.

## 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.
- 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.* 1: S1-S2.
- Trowsdale, J., Barten, R., Haude, A., Stewart, C.A., Beck, S. and Wilson M.J. 2001. The genomic context of natural killer receptor extended gene families. *Immunol. Rev.* 181: 20-38.
- Zumkeller, W., Volleth, M., Muschke, P., Tönnies, H., Heller, A., Liehr, T., Wieacker, P. and Stumm, M. 2004. Genotype/phenotype analysis in a patient with pure and complete trisomy 12p. *Am. J. Med. Genet. A* 129: 261-264.
- Kelley, J., Walter, L. and Trowsdale, J. 2005. Comparative genomics of natural killer cell receptor gene clusters. *PLoS Genet.* 1: e27.
- Nishimura, G., Haga, N., Kitoh, H., Tanaka, Y., Sonoda, T., Kitamura, M., Shirahama, S., Itoh, T., Nakashima, E., Ohashi, H. and Ikegawa, S. 2005. The phenotypic spectrum of COL2A1 mutations. *Hum. Mutat.* 26: 36-43.
- Segel, R., Peter, I., Demmer, L.A., Cowan, J.M., Hoffman, J.D. and Bianchi, D.W. 2006. The natural history of trisomy 12p. *Am. J. Med. Genet. A* 140: 695-703.
- Stein, R. 2007. Genetics of Noonan syndrome—a new gene, and the search is still on. *Clin. Genet.* 72: 402-404.
- van der Burgt, I. 2007. Noonan syndrome. *Orphanet J. Rare Dis.* 2: 4.

## CHROMOSOMAL LOCATION

Genetic locus: PARPBP (human) mapping to 12q23.2.

## SOURCE

C12orf48 (Y-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of C12orf48 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-245199 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

C12orf48 (Y-18) is recommended for detection of C12orf48 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 C12orf48 siRNA (h): sc-95807, C12orf48 shRNA Plasmid (h): sc-95807-SH and C12orf48 shRNA (h) Lentiviral Particles: sc-95807-V.

Molecular Weight of C12orf48 isoforms 1/2/3/4/5-7: 65/59/20/15/34 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.