SANTA CRUZ BIOTECHNOLOGY, INC.

C12orf50 (N-17): sc-242017

SANTA CRUZ BIOTECHNOLOGY

The Power to Question

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 C12orf50 gene product has been provisionally designated C12orf50 pending further characterization.

REFERENCES

1. 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.
2. 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.
3. Trowsdale, J., Barten, R., Haude, A., Stewart, C.A., Beck, S. et al. 2001. The genomic context of natural killer receptor extended gene families. Immunol. Rev. 181: 20-38.
4. 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.
5. Kelley, J., Walter, L. and Trowsdale, J. 2005. Comparative genomics of natural killer cell receptor gene clusters. PLoS Genet. 1: e27.
6. 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.
7. 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.
8. Stein, R. 2007. Genetics of Noonan syndrome-a new gene, and the search is still on. Clin. Genet. 72: 402-404.
9. van der Burgt, I. 2007. Noonan syndrome. Orphanet. J. Rare Dis. 2: 4.

CHROMOSOMAL LOCATION

Genetic locus: C12orf50 (human) mapping to 12q21.32.

STORAGE

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

SOURCE

C12orf50 (N-17) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of C12orf50 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-242017 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

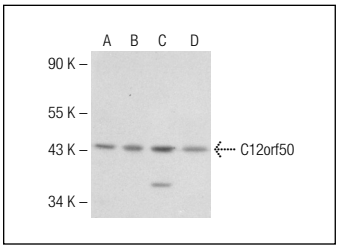
C12orf50 (N-17) is recommended for detection of C12orf50 of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for C12orf50 siRNA (h): sc-96078, C12orf50 shRNA Plasmid (h): sc-96078-SH and C12orf50 shRNA (h) Lentiviral Particles: sc-96078-V.

Molecular Weight of C12orf50: 47 kDa.

Positive Controls: Hep G2 cell lysate: sc-2227, Jurkat whole cell lysate: sc-2204 or DU 145 cell lysate: sc-2268.

DATA



C12orf50 (N-17): sc-242017. Western blot analysis of C12orf50 expression in Hep G2 (**A**), Jurkat (**B**), DU 145 (**C**) and HEL 92.1.7 (**D**) whole cell lysates.

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.