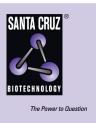
SANTA CRUZ BIOTECHNOLOGY, INC.

C12orf27 (S-15): sc-323542



BACKGROUND

C12orf27 (chromosome 12 open reading frame 27), also known as NCRNA00262 (non-protein coding RNA 262) or FLJ38690, is a 134 amino acid protein that is encoded by a gene located on human chromosome 12, which encodes over 1,100 genes and comprises approximately 4.5% of the human genome. Chromosome 12 is associated with a number of skeletal deformities, 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. 1: S1-S2.
- 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 129: 261-264.
- Kelley, J., et al. 2005. Comparative genomics of natural killer cell receptor gene clusters. PLoS Genet. 1: e27.
- 6. Nishimura, G., et al. 2005. The phenotypic spectrum of COL2A1 mutations. Hum. Mutat. 26: 36-43.
- 7. Segel, R., et al. 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: HNF1A-AS1 (human) mapping to 12q24.23.

SOURCE

C12orf27 (S-15) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of C12orf27 of human origin.

STORAGE

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

PRODUCT

Each vial contains 200 μg lgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-323542 P, (100 μg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

C12orf27 (S-15) is recommended for detection of C12orf27 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 C12orf27 siRNA (h): sc-106864, C12orf27 shRNA Plasmid (h): sc-106864-SH and C12orf27 shRNA (h) Lentiviral Particles: sc-106864-V.

Molecular Weight of C12orf27: 17 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) 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™ 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.