

LOC645277 (S-15): sc-247721

BACKGROUND

LOC645277 is a 139 amino acid protein encoded by a gene that maps to human chromosome 12q24.33. 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 which vary in severity depending on the extent of mosaicism. It 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.
- Delgado Carrasco, J., et al. 2001. Achondrogenesis type II-hypochondrogenesis: radiological features. Case report. *An. Esp. Pediatr.* 55: 553-557.
- Yokoyama, T., et al. 2003. A case of Kniest dysplasia with retinal detachment and the mutation analysis. *Am. J. Ophthalmol.* 136: 1186-1188.
- Forzano, F., et al. 2007. A familial case of achondrogenesis type II caused by a dominant COL2A1 mutation and "patchy" expression in the mosaic father. *Am. J. Med. Genet. A* 143A: 2815-2820.
- Wainwright, H. and Beighton, P. 2008. Visceral manifestations of hypochondrogenesis. *Virchows Arch.* 453: 203-207.
- Lo, F.S., et al. 2009. High resolution melting analysis for mutation detection for PTPN11 gene: applications of this method for diagnosis of Noonan syndrome. *Clin. Chim. Acta* 409: 75-77.
- Benussi, D.G., et al. 2009. Trisomy 12p and monosomy 4p: phenotype-genotype correlation. *Genet. Test. Mol. Biomarkers* 13: 199-204.

CHROMOSOMAL LOCATION

Genetic locus: LRCOL1 (human) mapping to 12q24.33.

SOURCE

LOC645277 (S-15) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of LOC645277 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-247721 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

RESEARCH USE

For research use only, not for use in diagnostic procedures.

APPLICATIONS

LOC645277 (S-15) is recommended for detection of LOC645277 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 LOC645277 siRNA (h): sc-96035, LOC645277 shRNA Plasmid (h): sc-96035-SH and LOC645277 shRNA (h) Lentiviral Particles: sc-96035-V.

Molecular Weight of LOC645277: 16 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.

PROTOCOLS

See our web site at www.scbt.com or our catalog for detailed protocols and support products.