# SANTA CRUZ BIOTECHNOLOGY, INC.

# LOC100130172 (D-18): sc-247493



### BACKGROUND

LOC100130172 is a 173 amino acid protein that is encoded by a gene that maps to human chromosome 5. Chromosome 5 contains 181 million base pairs and comprises nearly 6% of the human genome. Chromosome 5 is associated with Cockayne syndrome through the ERCC8 gene and familial adenomatous polyposis through the adenomatous polyposis coli (APC) tumor suppressor gene. Treacher Collins syndrome is also chromosome 5-associated and is caused by insertions or deletions within the TCOF1 gene. Deletion of the p arm of chromosome 5 leads to cri du chat syndrome, while deletion of the q arm or of chromosome 5 altogether is common in therapy-related acute myelogenous leukemias and myelodysplastic syndrome.

## REFERENCES

- Edwards, S.J., et al. 1997. The mutational spectrum in Treacher Collins syndrome reveals a predominance of mutations that create a prematuretermination codon. Am. J. Hum. Genet. 60: 515-524.
- McDaniel, L.D., et al. 1997. Confirmation of homozygosity for a single nucleotide substitution mutation in a Cockayne syndrome patient using monoallelic mutation analysis in somatic cell hybrids. Hum. Mutat. 10: 317-321.
- Crawford, M.J., et al. 1997. Human and murine PTX1/Ptx1 gene maps to the region for Treacher Collins syndrome. Mamm. Genome 8: 841-845.
- Finch, R., et al. 2005. Familial adenomatous polyposis and mental retardation caused by a *de novo* chromosomal deletion at 5q15-q22: report of a case. Dis. Colon Rectum. 48: 2148-2152.
- Anindya, R., et al. 2007. Damage-induced ubiquitylation of human RNA polymerase II by the ubiquitin ligase Nedd4, but not Cockayne syndrome proteins or BRCA1. Mol. Cell 28: 386-397.
- Vera-Carbonell, A., et al. 2009. Characterization of a *de novo* complex chromosomal rearrangement in a patient with cri-du-chat and trisomy 5p syndromes. Am. J. Med. Genet. A 149A: 2513-2521.
- Ravandi, F., et al. 2009. Superior outcome with hypomethylating therapy in patients with acute myeloid leukemia and high-risk myelodysplastic syndrome and chromosome 5 and 7 abnormalities. Cancer 115: 5746-5751.
- 8. Sazawal, S., et al. 2009. Haematological & molecular profile of acute myelogenous leukaemia in India. Indian J. Med. Res. 129: 256-261.

#### CHROMOSOMAL LOCATION

Genetic locus: LOC100130172 (human) mapping to 5q31.2.

#### SOURCE

LOC100130172 (D-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of LOC100130172 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  $\mu g$  lgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-247493 P, (100  $\mu g$  peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

#### **APPLICATIONS**

L0C100130172 (D-18) is recommended for detection of L0C100130172 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).

Molecular Weight of LOC100130172: 22 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.