

# LOC100130976 (T-20): sc-247499

## BACKGROUND

LOC100130976 encoded by a gene that is located on chromosome 15. Encoding more than 700 genes, chromosome 15 is made up of approximately 106 million base pairs and is about 3% of the human genome. Angelman and Prader-Willi syndromes are associated with loss of function or deletion of genes in the 15q11-q13 region. In the case of Angelman syndrome, this loss is due to inactivity of the maternal 15q11-q13 encoded UBE3A gene in the brain by either chromosomal deletion or mutation. In cases of Prader-Willi syndrome, there is a partial or complete deletion of this region from the paternal copy of chromosome 15. Tay-Sachs disease is a lethal disorder associated with mutations of the HEXA gene, which is encoded by chromosome 15. Marfan syndrome is associated with chromosome 15 through the FBN1 gene.

## REFERENCES

1. Cachón-González, M.B., et al. 2006. Effective gene therapy in an authentic model of Tay-Sachs-related diseases. *Proc. Natl. Acad. Sci. USA* 103: 10373-10378.
2. Zody, M.C., et al. 2006. Analysis of the DNA sequence and duplication history of human chromosome 15. *Nature* 440: 671-675.
3. Diene, G., et al. 2007. The Prader-Willi syndrome. *Ann. Endocrinol.* 68: 129-137.
4. Lalande, M. and Calciano, M.A. 2007. Molecular epigenetics of Angelman syndrome. *Cell. Mol. Life Sci.* 64: 947-960.
5. Maegawa, G.H., et al. 2007. Pyrimethamine as a potential pharmacological chaperone for late-onset forms of GM2 gangliosidosis. *J. Biol. Chem.* 282: 9150-9161.
6. Makoff, A.J. and Flomen, R.H. 2007. Detailed analysis of 15q11-q14 sequence corrects errors and gaps in the public access sequence to fully reveal large segmental duplications at breakpoints for Prader-Willi, Angelman, and inv dup(15) syndromes. *Genome Biol.* 8: R114
7. Ramirez, F. and Dietz, H.C. 2007. Fibrillin-rich microfibrils: Structural determinants of morphogenetic and homeostatic events. *J. Cell. Physiol.* 213: 326-330.
8. ten Dijke, P. and Arthur, H.M. 2007. Extracellular control of TGF $\beta$  signalling in vascular development and disease. *Nat. Rev. Mol. Cell Biol.* 8: 857-869.

## CHROMOSOMAL LOCATION

Genetic locus: LOC100130976 (human) mapping to 15q26.2.

## SOURCE

LOC100130976 (T-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of LOC100130976 of human origin.

## STORAGE

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

## PRODUCT

Each vial contains 200  $\mu$ g IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-247499 P, (100  $\mu$ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

LOC100130976 (T-20) is recommended for detection of LOC100130976 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 LOC100130976: 14 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.

## 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.