LOC100132292 (S-20): sc-247518



The Power to Question

BACKGROUND

LOC100132292 is a 168 amino acid protein that is encoded by a gene that is located on chromosome 15q11.2. 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

- 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.
- 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.
- 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
- 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 β signalling in vascular development and disease. Nat. Rev. Mol. Cell Biol. 8: 857-869.

CHROMOSOMAL LOCATION

Genetic locus: LOC100132292 (human) mapping to 15q11.2.

SOURCE

LOC100132292 (S-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of LOC100132292 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 μg IgG in 1.0 ml of PBS with <0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-247518 P, (100 μ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

LOC100132292 (S-20) is recommended for detection of LOC100132292 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 LOC100132292: 18 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 or our catalog for detailed protocols and support products.

Santa Cruz Biotechnology, Inc. 1.800.457.3801 831.457.3801 **Europe** +00800 4573 8000 49 6221 4503 0 **www.scbt.com**