

C9orf53 (A-18): sc-323703

BACKGROUND

Chromosome 9 consists of about 145 million bases and 4% of the human genome and encodes nearly 900 genes. Considered to play a role in gender determination, deletion of the distal portion of 9p can lead to development of male to female sex reversal, the phenotype of a female with a male X,Y genotype. Hereditary hemorrhagic telangiectasia, which is characterized by harmful vascular defects, is associated with the chromosome 9 gene encoding endoglin protein, ENG. Familial dysautonomia is also associated with chromosome 9 through the gene IKBKAP. Notably, chromosome 9 encompasses the largest interferon family gene cluster. Chromosome 9 is partnered with chromosome 22 in the translocation leading to the aberrant production of Bcr-Abl fusion protein often found in leukemias. The C9orf53 gene product has been provisionally designated C9orf53 pending further characterization.

REFERENCES

- Humphray, S.J., et al. 2004. DNA sequence and analysis of human chromosome 9. *Nature* 429: 369-374.
- Zheng, X., et al. 2006. BCR and its mutants, the reciprocal t(9;22)-associated ABL/BCR fusion proteins, differentially regulate the cytoskeleton and cell motility. *BMC Cancer* 6: 262.
- Coppo, P., et al. 2006. BCR-ABL activates STAT3 via JAK and MEK pathways in human cells. *Br. J. Haematol.* 134: 171-179.
- Hims, M.M., et al. 2007. A humanized IKBKAP transgenic mouse models a tissue-specific human splicing defect. *Genomics* 90: 389-396.
- Burmeister, T., et al. 2007. Atypical BCR-ABL mRNA transcripts in adult acute lymphoblastic leukemia. *Haematologica* 92: 1699-1702.
- Fernandez-L, A., et al. 2007. Gene expression fingerprinting for human hereditary hemorrhagic telangiectasia. *Hum. Mol. Genet.* 16: 1515-1533.
- Cottin, V., et al. 2007. Pulmonary vascular manifestations of hereditary hemorrhagic telangiectasia (Rendu-Osler disease). *Respiration* 74: 361-378.
- Gardiner, J., et al. 2007. Potential role of tubulin acetylation and microtubule-based protein trafficking in familial dysautonomia. *Traffic* 8: 1145-1149.

CHROMOSOMAL LOCATION

Genetic locus: C9orf53 (human) mapping to 9p21.3.

SOURCE

C9orf53 (A-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of C9orf53 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.

PROTOCOLS

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

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-323703 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

C9orf53 (A-18) is recommended for detection of C9orf53 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 C9orf53 siRNA (h): sc-92894, C9orf53 shRNA Plasmid (h): sc-92894-SH and C9orf53 shRNA (h) Lentiviral Particles: sc-92894-V.

Molecular Weight of C9orf53: 9 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.