

# C9orf50 (D-19): sc-246022

## 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 C9orf50 gene product has been provisionally designated C9orf50 pending further characterization.

## REFERENCES

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3. Coppo, P., et al. 2006. Bcr-Abl activates STAT3 via JAK and MEK pathways in human cells. *Br. J. Haematol.* 134: 171-179.
4. Hims, M.M., et al. 2007. A humanized IKBKAP transgenic mouse models a tissue-specific human splicing defect. *Genomics* 90: 389-396.
5. Burmeister, T., et al. 2007. Atypical Bcr-Abl mRNA transcripts in adult acute lymphoblastic leukemia. *Haematologica* 92: 1699-1702.
6. Fernandez-L, A., et al. 2007. Gene expression fingerprinting for human hereditary hemorrhagic telangiectasia. *Hum. Mol. Genet.* 16: 1515-1533.
7. Cottin, V., et al. 2007. Pulmonary vascular manifestations of hereditary hemorrhagic telangiectasia (Rendu-Osler disease). *Respiration* 74: 361-378.
8. Gardiner, J., et al. 2007. Potential role of tubulin acetylation and microtubule-based protein trafficking in familial dysautonomia. *Traffic* 8: 1145-1149.
9. Temtamy, S.A., et al. 2007. Phenotypic and cytogenetic spectrum of 9p trisomy. *Genet. Couns.* 18: 29-48.

## CHROMOSOMAL LOCATION

Genetic locus: C9orf50 (human) mapping to 9q34.11.

## SOURCE

C9orf50 (D-19) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the C-terminus of C9orf50 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-246022 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

C9orf50 (D-19) is recommended for detection of C9orf50 of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], 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 C9orf50 siRNA (h): sc-92936, C9orf50 shRNA Plasmid (h): sc-92936-SH and C9orf50 shRNA (h) Lentiviral Particles: sc-92936-V.

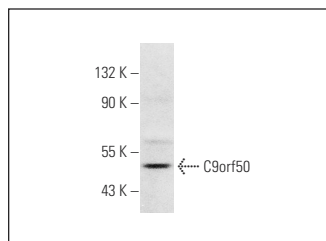
Molecular Weight of C9orf50: 48 kDa.

Positive Controls: human kidney extract: sc-363764.

## 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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) 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.

## DATA



C9orf50 (D-19): sc-246022. Western blot analysis of C9orf50 expression in human kidney tissue extract.

## 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.