

Torsin4A (L-17): sc-246006

BACKGROUND

C9orf167 (chromosome 9 open reading frame 167) is a 423 amino acid single-pass membrane protein that belongs to the clpA/clpB family and torsin sub-family. The gene encoding C9orf167 maps to human chromosome 9q34.3. Chromosome 9 consists of about 145 million bases, represents 4% of the human genome and encodes nearly 900 genes. Thought 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 though 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.

REFERENCES

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3. 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* 7: 262.
4. Burmeister, T., et al. 2007. Atypical Bcr-Abl mRNA transcripts in adult acute lymphoblastic leukemia. *Haematologica* 92: 1699-1702.
5. Cottin, V., et al. 2007. Pulmonary vascular manifestations of hereditary hemorrhagic telangiectasia (Rendu-Osler disease). *Respiration* 74: 361-378.
6. Fernandez-L, A., et al. 2007. Gene expression fingerprinting for human hereditary hemorrhagic telangiectasia. *Hum. Mol. Genet.* 16: 1515-1533.
7. Gardiner, J., et al. 2007. Potential role of tubulin acetylation and microtubule-based protein trafficking in familial dysautonomia. *Traffic* 8: 1145-1149.
8. Hims, M.M., et al. 2007. A humanized IKBKAP transgenic mouse models a tissue-specific human splicing defect. *Genomics* 90: 389-396.

CHROMOSOMAL LOCATION

Genetic locus: TOR4A (human) mapping to 9q34.3; Tor4a (mouse) mapping to 2 A3.

SOURCE

Torsin4A (L-17) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of Torsin4A 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-246006 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

Torsin4A (L-17) is recommended for detection of Torsin4A of mouse, rat and 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).

Torsin4A (L-17) is also recommended for detection of Torsin4A in additional species, including equine, canine and porcine.

Suitable for use as control antibody for Torsin4A siRNA (h): sc-92969, Torsin4A siRNA (m): sc-141953, Torsin4A shRNA Plasmid (h): sc-92969-SH, Torsin4A shRNA Plasmid (m): sc-141953-SH, Torsin4A shRNA (h) Lentiviral Particles: sc-92969-V and Torsin4A shRNA (m) Lentiviral Particles: sc-141953-V.

Molecular Weight of Torsin4A: 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.

PROTOCOLS

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