



TBC1D22A (h3): 293 Lysate: sc-114524

BACKGROUND

GTPase-activating proteins (GAPs) accelerate the intrinsic rate of GTP hydrolysis of Ras-related proteins, resulting in downregulation of their active form. TBC1D22A (TBC1 domain family, member 22A), also known as C22orf4, is a 517 amino acid protein that contains one Rab-GAP TBC domain and is thought to function as a GTPase-activating protein for Rab family members. Multiple isoforms of TBC1D22A exist due to alternative splicing events. The gene encoding TBC1D22A maps to human chromosome 22, which houses over 500 genes and is the second smallest human chromosome. Mutations in several of the genes that map to chromosome 22 are involved in the development of Phelan-McDermid syndrome, neurofibromatosis type 2, autism and schizophrenia. Additionally, translocations between chromosomes 9 and 22 may lead to the formation of the Philadelphia chromosome and the subsequent production of the novel fusion protein BCR-Abl, a potent cell proliferation activator found in several types of leukemias.

REFERENCES

1. Gilbert, F. 1998. Disease genes and chromosomes: disease maps of the human genome. Chromosome 22. Genet. Test. 2: 89-97.
2. Schwab, S.G. and Wildenauer, D.B. 1999. Chromosome 22 workshop report. Am. J. Med. Genet. 88: 276-278.
3. Arinami, T. 2006. Analyses of the associations between the genes of 22q11 deletion syndrome and schizophrenia. J. Hum. Genet. 51: 1037-1045.
4. Hay, B.N. 2007. Deletion 22q11: spectrum of associated disorders. Semin. Pediatr. Neurol. 14: 136-139.
5. Tsilchorozidou, T., Menko, F.H., Laloo, F., Kidd, A., De Silva, R., Thomas, H., Smith, P., Malcolmson, A., Dore, J., Madan, K., Brown, A., Yovos, J.G., Tsaligopoulos, M., Vogiatzis, N., Baser, M.E., Wallace, A.J. and Evans, D.G. 2004. Constitutional rearrangements of chromosome 22 as a cause of neurofibromatosis 2. J. Med. Genet. 41: 529-534.
6. Tempel, W., Tong, Y., Dimov, S., Bochkarev, A. and Park, H. 2008. First crystallographic models of human TBC domains in the context of a family-wide structural analysis. Proteins 71: 497-502.
7. Ishibashi, K., Kanno, E., Itoh, T. and Fukuda, M. 2009. Identification and characterization of a novel Tre-2/Bub2/Cdc16 (TBC) protein that possesses Rab3A-GAP activity. Genes Cells 14: 41-52.

CHROMOSOMAL LOCATION

Genetic locus: TBC1D22A (human) mapping to 22q13.31.

PRODUCT

TBC1D22A (h3): 293 Lysate represents a lysate of human TBC1D22A transfected 293 cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

APPLICATIONS

TBC1D22A (h3): 293 Lysate is suitable as a Western Blotting positive control for human reactive TBC1D22A antibodies. Recommended use: 10-20 µl per lane.

Control 293 Lysate: sc-110760 is available as a Western Blotting negative control lysate derived from non-transfected 293 cells.

RESEARCH USE

For research use only, not for use in diagnostic procedures.

PROTOCOLS

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