



## ENTHD1 (S-18): sc-86434

### BACKGROUND

Chromosome 22 contains over 500 genes and about 49 million bases. Being the second smallest human chromosome, 22 contains a surprising variety of interesting genes. Phelan-McDermid syndrome, neurofibromatosis type 2 and autism are associated with chromosome 22. A schizophrenia susceptibility locus has been identified on chromosome 22 and studies show that 22q11 deletion symptoms include a high incidence of schizophrenia. 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 leukemia.

### REFERENCES

- Gilbert, F. 1998. Disease genes and chromosomes: disease maps of the human genome. *Chromosome 22. Genet. Test.* 2: 89-97.
- Schwab, S.G. and Wildenauer, D.B. 1999. Chromosome 22 workshop report. *Am. J. Med. Genet.* 88: 276-278.
- 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.
- Arinami, T. 2006. Analyses of the associations between the genes of 22q11 deletion syndrome and schizophrenia. *J. Hum. Genet.* 51: 1037-1045.
- Paylor, R., Glaser, B., Mupo, A., Ataliotis, P., Spencer, C., Sobotka, A., Sparks, C., Choi, C.H., Oghalai, J., Curran, S., Murphy, K.C., Monks, S., Williams, N., O'Donovan, M.C., Owen, M.J., Scambler, P.J. and Lindsay, E. 2006. TBX1 haploinsufficiency is linked to behavioral disorders in mice and humans: implications for 22q11 deletion syndrome. *Proc. Natl. Acad. Sci. USA* 103: 7729-7734.
- Zheng, X., Güller, S., Beissert, T., Puccetti, E. and Ruthardt, M. 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.
- Ahronowitz, I., Xin, W., Kiely, R., Sims, K., MacCollin, M. and Nunes, F.P. 2007. Mutational spectrum of the NF2 gene: a meta-analysis of 12 years of research and diagnostic laboratory findings. *Hum. Mutat.* 28: 1-12.
- Hay, B.N. 2007. Deletion 22q11: spectrum of associated disorders. *Semin. Pediatr. Neurol.* 14: 136-139.

### CHROMOSOMAL LOCATION

Genetic locus: ENTHD1 (human) mapping to 22q13.1.

### SOURCE

ENTHD1 (S-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of ENTHD1 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-86434 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

### APPLICATIONS

ENTHD1 (S-18) is recommended for detection of ENTHD1 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 ENTHD1 siRNA (h): sc-77276, ENTHD1 shRNA Plasmid (h): sc-77276-SH and ENTHD1 shRNA (h) Lentiviral Particles: sc-77276-V.

### 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](http://www.scbt.com) or our catalog for detailed protocols and support products.