

TTLL1 (h4): 293T Lysate: sc-172036

BACKGROUND

A large protein group known as the tubulin tyrosine ligase-like (TTLL) family is implied to catalyze ligations of amino acids to tubulins and other substrates. Each member contains a characteristic TTL domain. TTLL1 (Tubulin tyrosine ligase-like family, member 1), also known as tubulin polyglutamylase complex subunit 3, PGs3 or C22orf7, is a 423 amino acid catalytic subunit of the neuronal tubulin polyglutamylase complex and a member of the tubulin polyglutamylase family. Localized to cytoskeleton and cytosol, TTLL1 is widely expressed with highest levels found in brain, testis and heart. TTLL1 generates glutamate side chains on C-terminal regions of α - and β -Tubulin and contains one TTL domain. Four TTLL1 isoforms are known to exist as a result of alternative splicing events. The gene encoding TTLL1 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.

REFERENCES

1. Trichet, V., et al. 2000. Characterization of the human tubulin tyrosine ligase-like 1 gene (TTLL1) mapping to 22q13.1. *Gene* 257: 109-117.
2. Briegel, W. and Cohen, M. 2004. Chromosome 22q11 deletion syndrome and its relevance for child and adolescent psychiatry. An overview of etiology, physical symptoms, aspects of child development and psychiatric disorders. *Z. Kinder Jugendpsychiatr. Psychother.* 32: 107-115.
3. Janke, C., et al. 2005. Tubulin polyglutamylase enzymes are members of the TTL domain protein family. *Science* 308: 1758-1762.
4. Online Mendelian Inheritance in Man, OMIM™. 2007. Johns Hopkins University, Baltimore, MD. MIM Number: 608955. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
5. Gothelf, D., et al. 2008. Genes, brain development and psychiatric phenotypes in velo-cardio-facial syndrome. *Dev. Disabil. Res. Rev.* 14: 59-68.
6. Sathyamoorthi, S., et al. 2009. Array analysis and molecular studies of INI1 in an infant with deletion 22q13 (Phelan-McDermid syndrome) and atypical teratoid/rhabdoid tumor. *Am. J. Med. Genet. A* 149A: 1067-1069.
7. Vorstman, J.A., et al. 2009. Association of the PIK4CA schizophrenia-susceptibility gene in adults with the 22q11.2 deletion syndrome. *Am. J. Med. Genet. B Neuropsychiatr. Genet.* 150B: 430-433.
8. Rogowski, K., et al. 2009. Evolutionary divergence of enzymatic mechanisms for posttranslational polyglutamylation. *Cell* 137: 1076-1087.
9. Evans, D.G. 2009. Neurofibromatosis 2 (bilateral acoustic neurofibromatosis, central neurofibromatosis, NF2, neurofibromatosis type II). *Genet. Med.* 11: 599-610.

CHROMOSOMAL LOCATION

Genetic locus: TTLL1 (human) mapping to 22q13.2.

PRODUCT

TTLL1 (h4): 293T Lysate represents a lysate of human TTLL1 transfected 293T cells and is provided as 100 μ g protein in 200 μ l SDS-PAGE buffer.

APPLICATIONS

TTLL1 (h4): 293T Lysate is suitable as a Western Blotting positive control for human reactive TTLL1 antibodies. Recommended use: 10-20 μ l per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

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.

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.