

PUS1 (h2): 293T Lysate: sc-172003

BACKGROUND

PUS1 (psuedouridine synthase 1) belongs to the tRNA pseudouridine synthase truA family. PUS1 functions in the conversion of uridine into pseudouridine after the nucleotide has been incorporated into RNA. It may have a functional role in tRNAs and is also thought to assist in the peptidyl transfer reaction of rRNAs. As a nucleus-resident protein, PUS1 forms a complex with RARG and the SRA1 RNA. PUS1 is widely expressed, with highest levels of expression in the brain and skeletal muscle tissues. Defects in PUS1 are a cause of myopathy with lactic acidosis and sideroblastic anemia (MLASA), also known as mitochondrial myopathy and sideroblastic anemia. MLASA is a rare autosomal recessive oxidative phosphorylation disorder specific to bone marrow and skeletal muscle. The deduced human PUS1 protein contains 348 amino acids and shares 92% sequence homology with mouse PUS1.

REFERENCES

1. Arluison, V., Hountondji, C., Robert, B. and Grosjean, H. 1998. Transfer RNA-pseudouridine synthetase Pus1 of *Saccharomyces cerevisiae* contains one atom of zinc essential for its native conformation and tRNA recognition. *Biochemistry* 37: 7268-7276.
2. Arluison, V., Batelier, G., Riès-Kautt, M. and Grosjean, H. 1999. RNA: pseudouridine synthetase Pus1 from *Saccharomyces cerevisiae*: oligomerization property and stoichiometry of the complex with yeast tRNA(Phe). *Biochimie* 81: 751-756.
3. Arluison, V., Buckle, M. and Grosjean, H. 1999. Pseudouridine synthetase Pus1 of *Saccharomyces cerevisiae*: kinetic characterisation, tRNA structural requirement and real-time analysis of its complex with tRNA. *J. Mol. Biol.* 289: 491-502.
4. Chen, J. and Patton, J.R. 1999. Cloning and characterization of a mammalian pseudouridine synthase. *RNA* 5: 409-419.
5. Chen, J. and Patton, J.R. 2001. Mouse pseudouridine synthase 1: gene structure and alternative splicing of pre-mRNA. *Biochem. J.* 352: 465-473.
6. Online Mendelian Inheritance in Man, OMIM™. 2002. Johns Hopkins University, Baltimore, MD. MIM Number: 608109. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
7. Patton, J.R. and Padgett, R.W. 2003. *Caenorhabditis elegans* pseudouridine synthase 1 activity *in vivo*: tRNA is a substrate, but not U2 small nuclear RNA. *Biochem. J.* 372: 595-602.
8. Bykhovskaya, Y., Casas, K., Mengesha, E., Inbal, A. and Fischel-Ghodsian, N. 2004. Missense mutation in pseudouridine synthase 1 (PUS1) causes mitochondrial myopathy and sideroblastic anemia (MLASA). *Am. J. Hum. Genet.* 74: 1303-1308.
9. Patton, J.R., Bykhovskaya, Y., Mengesha, E., Bertolotto, C. and Fischel-Ghodsian, N. 2005. Mitochondrial myopathy and sideroblastic anemia (MLASA): missense mutation in the pseudouridine synthase 1 (PUS1) gene is associated with the loss of tRNA pseudouridylation. *J. Biol. Chem.* 280: 19823-19828.

CHROMOSOMAL LOCATION

Genetic locus: PUS1 (human) mapping to 12q24.33.

PRODUCT

PUS1 (h2): 293T Lysate represents a lysate of human PUS1 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

PUS1 (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive PUS1 antibodies. Recommended use: 10-20 µl per lane.

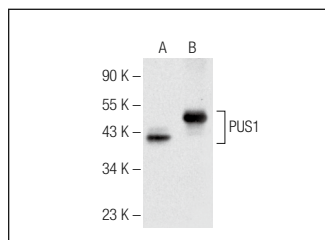
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

PUS1 (C-4): sc-376442 is recommended as a positive control antibody for Western Blot analysis of enhanced human PUS1 expression in PUS1 transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA



PUS1 (C-4): sc-376442. Western blot analysis of PUS1 expression in non-transfected: sc-117752 (A) and human PUS1 transfected: sc-172003 (B) 293T whole cell lysates.

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.