



Wolframin (P-13): sc-47939

BACKGROUND

The Wolframin gene encodes a protein found in endoplasmic reticulum membrane of several tissues including brain, pancreas, lung and placenta. Loss-of-function mutations in both alleles result in Wolfram syndrome (also known as DIDMOAD, an autosomal recessive disorder that causes juvenile diabetes mellitus, diabetes insipidus, optic atrophy and a number of neurological symptoms including deafness, ataxia and peripheral neuropathy. A large number and variety of mutations in this gene, particularly in exon 8, can be associated with Wolfram syndrome. Mutations in this gene can also cause autosomal dominant deafness 6 (DFNA6), also known as DFNA14 or DFNA38.

REFERENCES

- Osman, A.A., Saito, M., Makepeace, C., Permutt, M.A., Schlesinger, P. and Mueckler, M. 2003. Wolframin expression induces novel ion channel activity in endoplasmic reticulum membranes and increases intracellular calcium. *J. Biol. Chem.* 278: 52755-527562.
- Larsen, Z.M., Johannesen, J., Kristiansen, O.P., Nerup, J., Pociot, F., Danish IDDM Epidemiology and Genetics Group; Danish Study Group of IDDM in Childhood. 2004. Evidence for linkage on chromosome 4p16.1 in Type 1 diabetes Danish families and complete mutation scanning of the WFS1 (Wolframin) gene. *Diabet Med.* 21: 218-222.
- Smith, C.J., Crock, P.A., King, B.R., Meldrum, C.J. and Scott, R.J. 2004. Phenotype-genotype correlations in a series of Wolfram syndrome families. *Diabetes Care* 27: 2003-2009.
- Yamaguchi, S., Ishihara, H., Tamura, A., Yamada, T., Takahashi, R., Takei, D., Katagiri, H. and Oka, Y. 2004. Endoplasmic reticulum stress and N-glycosylation modulate protein. *Biochem. Biophys. Res. Commun.* 325: 250-256.
- Fonseca, S.G., Fukuma, M., Lipson, K.L., Nguyen, L.X., Allen, J.R., Oka, Y. and Urano, F. 2005. WFS1 is a novel component of the unfolded protein response and maintains homeostasis of the endoplasmic reticulum in pancreatic β -cells. *J. Biol. Chem.* 280: 39609-39615.
- Koido, K., Kōks, S., Nikopensius, T., Maron, E., Altmäe, S., Heinaste, E., Vabrit, K., Tammekivi, V., Hallast, P., Kurg, A., Shlik, J., Vasar, V., Metspalu, A. and Vasar, E. 2005. Polymorphisms in Wolframin (WFS1) gene are possibly related to for mood disorders. *Int. J. Neuropsychopharmacol.* 8: 235-244.
- Philbrook, C., Fritz, E. and Weiher, H. 2005. Expressional and functional studies of Wolframin, the gene function deficient in Wolfram syndrome, in mice and patient cells. *Exp. Gerontol.* 40: 671-678.
- Swift, M. and Swift, R.G. 2005. Wolframin mutations and hospitalization for psychiatric illness. *Mol. Psychiatry* 10: 799-803.
- Domenech, E., Gomez-Zaera, M. and Nunes, V. 2006. Wolfram/DIDMOAD syndrome, a heterogenic and molecularly complex neurodegenerative disease. *Pediatr. Endocrinol. Rev.* 3: 249-257.

CHROMOSOMAL LOCATION

Genetic locus: WFS1 (human) mapping to 4p16; Wfs1 (mouse) mapping to 5 B3.

SOURCE

Wolframin (P-13) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of Wolframin of human origin.

PRODUCT

Each vial contains 200 μ g IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, ready P, (100 μ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

Wolframin (P-13) is recommended for detection of Wolframin 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 Wolframin siRNA (h): sc-61804.

Molecular Weight of Wolframin: 100 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.

STORAGE

Store at 4° C, ****DO NOT FREEZE****. Stable for one year from the date of shipment. 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 or our catalog for detailed protocols and support products.