



PMM1 (h): 293T Lysate: sc-129495

BACKGROUND

PMM1 (phosphomannomutase 1), also known as Sec53 or PMMH22, is a 262 amino acid protein that localizes to the cytoplasm and belongs to the eukaryotic PMM family. Expressed at high levels in brain, liver, pancreas and heart and present at lower levels in skeletal muscle, PMM1 catalyzes the conversion of α -D-mannose 1-phosphate to D-mannose 6-phosphate, an event that is required for a number of crucial mannosyl transfer reactions. The gene encoding PMM1 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. Wada, Y. and Sakamoto, M. 1997. Isolation of the human phosphomannomutase gene (PMM1) and assignment to chromosome 22q13. *Genomics* 39: 416-417.
2. Matthijs, G., Schollen, E., Pirard, M., Budarf, M.L., Van Schaftingen, E. and Cassiman, J.J. 1997. PMM (PMM1), the human homologue of Sec53 or yeast phosphomannomutase, is localized on chromosome 22q13. *Genomics* 40: 41-47.
3. Hansen, S.H., Frank, S.R. and Casanova, J.E. 1997. Cloning and characterization of human phosphomannomutase, a mammalian homologue of yeast Sec53. *Glycobiology* 7: 829-834.
4. Gilbert, F. 1998. Disease genes and chromosomes: disease maps of the human genome. *Chromosome 22. Genet. Test.* 2: 89-97.
5. Schwab, S.G. and Wildenauer, D.B. 1999. Chromosome 22 workshop report. *Am. J. Med. Genet.* 88: 276-278.
6. Arinami, T. 2006. Analyses of the associations between the genes of 22q11 deletion syndrome and schizophrenia. *J. Hum. Genet.* 51: 1037-1045.
7. Online Mendelian Inheritance in Man, OMIM™. 2007. Johns Hopkins University, Baltimore, MD. MIM Number: 601786. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
8. Veiga-da-Cunha, M., Vleugels, W., Maliekal, P., Matthijs, G. and Van Schaftingen, E. 2008. Mammalian phosphomannomutase PMM1 is the brain IMP-sensitive glucose-1,6-bisphosphatase. *J. Biol. Chem.* 283: 33988-33993.

CHROMOSOMAL LOCATION

Genetic locus: PMM1 (human) mapping to 22q13.2.

PRODUCT

PMM1 (h): 293T Lysate represents a lysate of human PMM1 transfected 293T cells and is provided as 100 μ g protein in 200 μ l 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

PMM1 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive PMM1 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.

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.