



## PMM1 (C-19): sc-86200

### 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 alpha-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.
9. Romanowski, T., Sikorska, K. and Bielawski, K.P. 2008. GUS and PMM1 as suitable reference genes for gene expression analysis in the liver tissue of patients with chronic hepatitis. *Med. Sci. Monit.* 14: BR147-BR152.

### STORAGE

Store at 4° C, **\*\*DO NOT FREEZE\*\***. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

### PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) or our catalog for detailed protocols and support products.

### CHROMOSOMAL LOCATION

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

### SOURCE

PMM1 (C-19) is an affinity purified rabbit polyclonal antibody raised against a peptide mapping at the C-terminus of PMM1 of human origin.

### PRODUCT

Each vial contains 100 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-86200 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

### APPLICATIONS

PMM1 (C-19) is recommended for detection of PMM1 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 PMM1 siRNA (h): sc-76177, PMM1 shRNA Plasmid (h): sc-76177-SH and PMM1 shRNA (h) Lentiviral Particles: sc-76177-V.

Molecular Weight of PMM1: 30 kDa.

### RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (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 goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

### RESEARCH USE

For research use only, not for use in diagnostic procedures.