SANTA CRUZ BIOTECHNOLOGY, INC.

# PMM2 (AT3B4): sc-517420

## BACKGROUND

PMM2 (phosphomannomutase 2), also known as CDG1, CDGS or CDG1a, is a 246 amino acid protein that belongs to the eukaryotic PMM family. Localizing to cytoplasm, PMM2 encodes phosphomannomutase (PMM), an enzyme necessary for the synthesis of GDP-mannose. PMM2 mutations may cause defects in glycoprotein biosynthesis, which manifests as the congenital disorder carbohydrate-deficient glycoprotein syndrome type I. This syndrome is an autosomal recessive disorder characterized by severe encephalopathy with axial hypotonia, abnormal eye movement, pronounced psychomotor retardation, peripheral neuropathy, cerebellar hypoplasia and retinitis pigmentosa. Peculiar distribution of subcutaneous fat, nipple retraction and hypogonadism are also exhibited. The gene that encodes PMM2 maps to human chromosome 16p13.2.

## REFERENCES


1. Matthijs, G., et al. 1997. Mutations in PMM2, a phosphomannomutase gene on chromosome 16p13, in carbohydrate-deficient glycoprotein type I syndrome (Jaeken syndrome). Nat. Genet. 16: 88-92.
2. Kjaergaard, S., et al. 1998. Absence of homozygosity for predominant mutations in PMM2 in Danish patients with carbohydrate-deficient glycoprotein syndrome type 1. Eur. J. Hum. Genet. 6: 331-336.
3. Schollen, E., et al. 1998. Comparative analysis of the phosphomannomutase genes PMM1, PMM2 and PMM2psi: the sequence variation in the processed pseudogene is a reflection of the mutations found in the functional gene. Hum. Mol. Genet. 7: 157-164.
4. Kondo, I., et al. 1999. Missense mutations in phosphomannomutase 2 gene in two Japanese families with carbohydrate-deficient glycoprotein syndrome type 1. Clin. Genet. 55: 50-54.
5. Kjaergaard, S., et al. 1999. Carbohydrate-deficient glycoprotein syndrome type 1A: expression and characterisation of wild type and mutant PMM2 in *E. coli*. Eur. J. Hum. Genet. 7: 884-888.
6. Vuillaumier-Barrot, S., et al. 1999. Characterization of the 415G>A (E139K) PMM2 mutation in carbohydrate-deficient glycoprotein syndrome type Ia disrupting a splicing enhancer resulting in exon 5 skipping. Hum. Mutat. 14: 543-544.


## CHROMOSOMAL LOCATION

Genetic locus: PMM2 (human) mapping to 16p13.2.

## SOURCE

PMM2 (AT3B4) is a mouse monoclonal antibody raised against a recombinant protein corresponding to amino acids 1-246 of PMM2 of human origin.

## PRODUCT

Each vial contains 100 µg $IgG_{2a}$ kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

## RESEARCH USE

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

## APPLICATIONS

PMM2 (AT3B4) is recommended for detection of PMM2 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), flow cytometry (1 µg per 1 x $10^6$ cells) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for PMM2 siRNA (h): sc-93284, PMM2 shRNA Plasmid (h): sc-93284-SH and PMM2 shRNA (h) Lentiviral Particles: sc-93284-V.

Molecular Weight of PMM2: 28 kDa.

## 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, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

## 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 for detailed protocols and support products.