SANTA CRUZ BIOTECHNOLOGY, INC.

PMM2 (2A5): sc-517179

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The Power to Question

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

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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 PMM2ψ: 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.
7. Matthijs, G., et al. 2000. Mutations in PMM2 that cause congenital disorders of glycosylation, type Ia (CDG-Ia). Hum. Mutat. 16: 386-394.
8. Bjursell, C., et al. 2000. PMM2 mutation spectrum, including 10 novel mutations, in a large CDG type 1A family material with a focus on Scandinavian families. Hum. Mutat. 16: 395-400.
9. Le Bizec, C., et al. 2005. A new insight into PMM2 mutations in the French population. Hum. Mutat. 25: 504-505.

CHROMOSOMAL LOCATION

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

SOURCE

PMM2 (2A5) is a mouse monoclonal antibody raised against amino acids 1-246 representing full length PMM2 of human origin.

PRODUCT

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

APPLICATIONS

PMM2 (2A5) is recommended for detection of PMM2 of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)] 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.

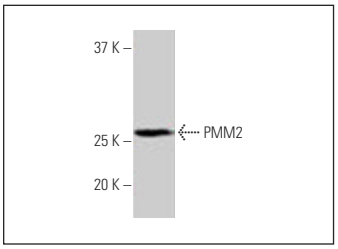
Molecular Weight of PMM2: 28 kDa.

Positive Controls: K-562 whole cell lysate: sc-2203.

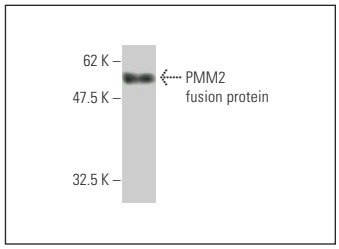
RECOMMENDED SUPPORT REAGENTS

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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml).

DATA



PMM2 (2A5): sc-517179. Western blot analysis of PMM2 expression in K-562 whole cell lysate.



PMM2 (2A5): sc-517179. Western blot analysis of human recombinant PMM2 fusion protein.

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 for detailed protocols and support products.