SANTA CRUZ BIOTECHNOLOGY, INC.

PMM2 siRNA (h): sc-93284

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.

PRODUCT

PMM2 siRNA (h) is a pool of 3 target-specific 19-25 nt siRNAs designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10 µM solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see PMM2 shRNA Plasmid (h): sc-93284-SH and PMM2 shRNA (h) Lentiviral Particles: sc-93284-V as alternate gene silencing products.

For independent verification of PMM2 (h) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-93284A, sc-93284B and sc-93284C.

STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20° C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20° C, avoid contact with RNAses and repeated freeze thaw cycles.

Resuspend lyophilized siRNA duplex in 330 µl of the RNAse-free water provided. Resuspension of the siRNA duplex in 330 µl of RNAse-free water makes a 10 µM solution in a 10 µM Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

APPLICATIONS

PMM2 siRNA (h) is recommended for the inhibition of PMM2 expression in human cells.

SUPPORT REAGENTS

For optimal siRNA transfection efficiency, Santa Cruz Biotechnology's siRNA Transfection Reagent: sc-29528 (0.3 ml), siRNA Transfection Medium: sc-36868 (20 ml) and siRNA Dilution Buffer: sc-29527 (1.5 ml) are recommended. Control siRNAs or Fluorescein Conjugated Control siRNAs are available as 10 µM in 66 µl. Each contain a scrambled sequence that will not lead to the specific degradation of any known cellular mRNA. Fluorescein Conjugated Control siRNAs include: sc-36869, sc-44239, sc-44240 and sc-44241. Control siRNAs include: sc-37007, sc-44230, sc-44231, sc-44232, sc-44233, sc-44234, sc-44235, sc-44236, sc-44237 and sc-44238.

GENE EXPRESSION MONITORING

PMM2 (2A5): sc-517179 is recommended as a control antibody for monitoring of PMM2 gene expression knockdown by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000) or immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500).

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, UltraCruz® Blocking Reagent: sc-516214 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.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor PMM2 gene expression knockdown using RT-PCR Primer: PMM2 (h)-PR: sc-93284-PR (20 µl). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

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.