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PMM2 Rabbit pAb

Catalog # AP97378

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	O15305
Other Accession	O15305
Reactivity	Human, Mouse
Predicted	Rat, Chicken, Dog, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	28082
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human PMM2
Epitope Specificity	1-100/246
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the eukaryotic PMM family.
DISEASE	Defects in PMM2 are the cause of congenital disorder of glycosylation type 1A (CDG1A) [MIM:212065]; also known as carbohydrate-deficient glycoprotein syndrome type Ia (CDGS1A) or Jaeken syndrome. Congenital disorders of glycosylation are metabolic deficiencies in glycoprotein biosynthesis that usually cause severe mental and psychomotor retardation. They are characterized by under-glycosylated serum glycoproteins. CDG1A is an autosomal recessive disorder characterized by a severe encephalopathy with axial hypotonia, abnormal eye movement, and pronounced psychomotor retardation, as well as peripheral neuropathy, cerebellar hypoplasia, and retinitis pigmentosa. Patients show a peculiar distribution of subcutaneous fat, nipple retraction, and hypogonadism.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	The protein encoded by this gene catalyzes the isomerization of mannose 6-phosphate to mannose 1-phosphate, which is a precursor to GDP-mannose necessary for the synthesis of dolichol-P-oligosaccharides. Mutations in this gene have been shown to cause defects in glycoprotein biosynthesis, which manifests as carbohydrate-deficient glycoprotein syndrome type I. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	5373
Other Names	CDG1; CDG1a; CDGS; PMI; PMI1; PMM 2; PMM2_BOVIN; PMM2; 5.4.2.8; PMM2_HUMAN; PMM2_MOUSE; phosphomannomutase 2; phosphomannose isomerase 1; mannose-6-phosphate isomerase; Congenital disorder of glycosylation, type Ia

Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Name	PMM2
Function	Involved in the synthesis of the GDP-mannose and dolichol-phosphate-mannose required for a number of critical mannosyl transfer reactions.
Cellular Location	Cytoplasm.

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.