

PMM2 Antibody (monoclonal) (M02)

Mouse monoclonal antibody raised against a full-length recombinant PMM2. Catalog # AT3356a

Product Information

Application WB
Primary Accession O15305
Other Accession BC008310
Reactivity Human
Host mouse
Clonality monoclonal
Isotype IgG2b Kappa

Clone Names 2A5 Calculated MW 28082

Additional Information

Gene ID 5373

Other Names Phosphomannomutase 2, PMM 2, PMM2

Target/Specificity PMM2 (AAH08310, 1 a.a. ~ 246 a.a) full-length recombinant protein with GST

tag. MW of the GST tag alone is 26 KDa.

Dilution WB~~1:500~1000

Format Clear, colorless solution in phosphate buffered saline, pH 7.2.

Storage Store at -20°C or lower. Aliquot to avoid repeated freezing and thawing.

Precautions PMM2 Antibody (monoclonal) (M02) is for research use only and not for use

in diagnostic or therapeutic procedures.

Background

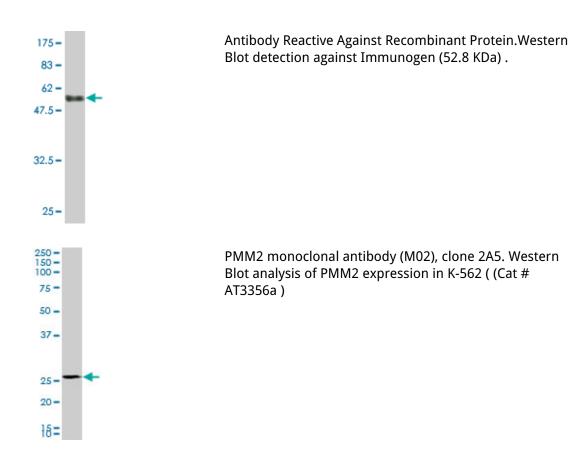
The protein encoded by this gene catalyzes the isomerization of mannose 6-phosphate to mannose 1-phosphate, which is a precursor to GDP-mannose necessary for the synthesis of dolichol-P-oligosaccharides. Mutations in this gene have been shown to cause defects in glycoprotein biosynthesis, which manifests as carbohydrate-deficient glycoprotein syndrome type I.

References

Functional analysis of three splicing mutations identified in the PMM2 gene: toward a new therapy for congenital disorder of glycosylation type Ia. Vega AI, et al. Hum Mutat, 2009 May. PMID 19235233.Primary skeletal dysplasia as a major manifesting feature in an infant with congenital disorder of glycosylation type

Ia. Coman D, et al. Am J Med Genet A, 2008 Feb 1. PMID 18203160. Cerebellar ataxia and congenital disorder of glycosylation Ia (CDG-Ia) with normal routine CDG screening. Vermeer S, et al. J Neurol, 2007 Oct. PMID 17694350. Congenital disorder of glycosylation type 1a: three siblings with a mild neurological phenotype. Coman D, et al. J Clin Neurosci, 2007 Jul. PMID 17451957. Characterization of two unusual truncating PMM2 mutations in two CDG-Ia patients. Schollen E, et al. Mol Genet Metab, 2007 Apr. PMID 17307006.

Images



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