

Recombinant human PMM2 protein

Catalog Number: ATGP0727

PRODUCT INFORMATION

Expression system

E.coli

Domain

1-246aa

UniProt No.

O15305

NCBI Accession No.

NP_000294.1

Alternative Names

Phosphomannomutase 2, CDG1, CDG1a, CDGS, Phosphomannomutase 2

PRODUCT SPECIFICATION

Molecular Weight

30.2 kDa (266aa) confirmed by MALDI-TOF

Concentration

1mg/ml (determined by Bradford assay)

Formulation

Liquid in. 20mM Tris-HCl buffer (pH 8.0) containing 10% glycerol, 1mM DTT, 0.1M NaCl.

Purity

> 90% by SDS-PAGE

Tag

His-Tag

Application

SDS-PAGE

Storage Condition

Can be stored at +2C to +8C for 1 week. For long term storage, aliquot and store at -20C to -80C. Avoid repeated freezing and thawing cycles.

BACKGROUND

Description

PMM2, also known as Phosphomannomutase 2, belongs to the eukaryotic PMM family. PMM2 is involved in the synthesis of the GDP-mannose and dolichol-phosphate-mannose required for a number of critical mannosyl transfer reactions. It catalyzes the isomerization of mannose 6-phosphate to mannose 1-phosphate. Mutations in PMM2 are associated with Congenital disorders of glycosylation (CDG) -Ia, an autosomal recessive disorder characterized by central nervous system dysfunction and multiorgan failure. Recombinant human PMM2 protein, fused to His-tag at N-terminus, was expressed in E. coli and purified by using conventional chromatography

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techniques.

Amino acid Sequence

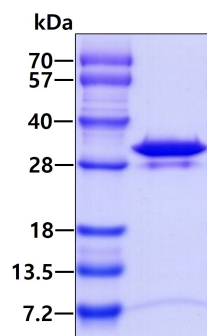
<MGSSHHHHHH SSGLVPRGSH> MAAPGPALCL FDVDGTLTAP RQKITKEMDD FLQKLRQKIK IGVVGGSDFE
KVQEQLGNDV VEKYDYVPE NGLVAYKDGK LLCRQNIQSH LGEALIQDLI NYCLSYIAKI KLPKKRGTFI EFRNGMLNVS
PIGRSCSQEE RIEFYELDKK ENIRQKFVAD LRKEFAGKGL TFSIGGQISF DVFPDGWDKR YCLRHVENDG YKTIYFFGDK
TMPGGNDHEI FTDPRTMGYS VTAPEDTRRI CELLFS

General References

Matthijs G., et al. (1997) Nat Genet. 16(1):88-92.
Vega Al., et al. (2009) Hum Mutat. 30(5):795-803.

DATA

SDS-PAGE



3 μ g by SDS-PAGE under reducing condition and visualized by coomassie blue stain.