abcam

Product datasheet

Recombinant Human PMM2 protein ab99391

1 Image

Overview

Product name	Recombinant Human PMM2 protein
Protein length	Full length protein

Description

Nature	Recombinant
Source	Escherichia coli

Amino Acid Sequence

Accession	O15305
Species	Human
Sequence	**MGSSHHHHHHSSGLVPRGSH**MAAPGPALCLFDVDGTLTAPRQKITKEMDD FLQKLRQKIKIGVVGGSDFEKVQEQLGNDVVEKYDYVFPENGLVAYKDGK LLCRQNIQSHLGEALIQDLINYCLSYIAKIKLPKKRGTFIEFRNGMLNVS PIGRSCSQEERIEFYELDKKENIRQKFVADLRKEFAGKGLTFSIGGQISF DVFPDGWDKRYCLRHVENDGYKTIYFFGDKTMPGGNDHEIFTDPRTMGYS VTAPEDTRRICELLFS
Molecular weight	30 kDa including tags
Amino acids	1 to 246
Tags	His tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab99391** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	SDS-PAGE Mass Spectrometry
Mass spectrometry	MALDI-TOF
Purity	> 95 % SDS-PAGE. ab99391 is purified using conventional chromatography techniques.
Form	Liquid

Preparation and Storage

Stability and Storage

Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.

Preservative: None
Constituents: 10% Glycerol, 0.1M Sodium chloride, 20mM Tris HCl, 1mM DTT, pH 8.0

General Info

Function

Involved in the synthesis of the GDP-mannose and dolichol-phosphate-mannose required for a number of critical mannosyl transfer reactions.

Pathway

Nucleotide-sugar biosynthesis; GDP-alpha-D-mannose biosynthesis; alpha-D-mannose 1-phosphate from D-fructose 6-phosphate: step 2/2.

Involvement in 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.

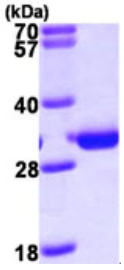
Sequence similarities

Belongs to the eukaryotic PMM family.

Cellular localization

Cytoplasm.

Images



SDS-PAGE - PMM2 protein (ab99391)

15% SDS-PAGE analysis of 3µg ab99391.

Our Abpromise to you: Quality guaranteed and expert technical support

- Replacement or refund for products not performing as stated on the datasheet
- Valid for 12 months from date of delivery
- Response to your inquiry within 24 hours

- We provide support in Chinese, English, French, German, Japanese and Spanish
- Extensive multi-media technical resources to help you
- We investigate all quality concerns to ensure our products perform to the highest standards

If the product does not perform as described on this datasheet, we will offer a refund or replacement. For full details of the Abpromise, please visit http://www.abcam.com/abpromise or contact our technical team.

Terms and conditions

- Guarantee only valid for products bought direct from Abcam or one of our authorized distributors