

Recombinant Human PMM2 protein ab99391

1 图像

描述	
产品名称	重组人PMM2蛋白
纯度	> 90 % SDS-PAGE. ab99391 is purified using conventional chromatography techniques.
表达系统	Escherichia coli
Accession	<u>O15305</u>
蛋白长度	Full length protein
无动物成分	No
性质	Recombinant
种属	Human
序列	MGSSHHHHHSSGLVPRGSHMAAPGPALCLFDVDGTLTAP RQKITKEMDD FLQKL RQKIKIGVVGGSDFEKVQEQLGNDVVEKYDYVFPENG LVAYKDGK LLCRQNIQSHLGEALIQDLINYLCLSYIAKIKLPKKRGTFIEF RNGMLNVS PIGRSCSQEERIEFYELDKKENIRQKFVADLRKEFAGKGLTF SIGGQISF DVFPDGWDKRYCLRHVENDGYKTIYFFGDKTMPGGNDHEIFT DPRTMGYS VTAPEDTRRICELLFS
预测分子量	30 kDa including tags
氨基酸	1 to 246
标签	His tag N-Terminus

技术指标	
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.	
应用	SDS-PAGE Mass Spectrometry
质谱法	MALDI-TOF
形式	Liquid

制备和贮存

稳定性和存储

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

pH: 8.00

Constituents: 0.0154% DTT, 0.316% Tris HCl, 10% Glycerol (glycerin, glycerine), 0.58% Sodium chloride

常规信息

功能

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

通路

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

疾病相关

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.

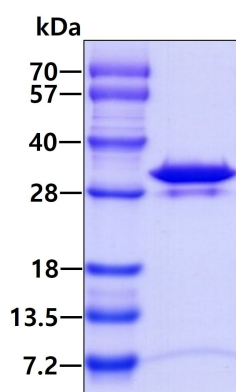
序列相似性

Belongs to the eukaryotic PMM family.

细胞定位

Cytoplasm.

图片



15% SDS-PAGE analysis of 3µg ab99391.

SDS-PAGE - Recombinant Human PMM2 protein
(ab99391)

Please note: All products are "FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC PROCEDURES"

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