abcam

Product datasheet

Recombinant Human POMGNT1 protein ab153784

Overview

Product name	Recombinant Human POMGNT1 protein
Protein length	Full length protein

Description

Nature	Recombinant
Source	HEK 293 cells

Amino Acid Sequence

Accession	Q8WZA1
Species	Human
Sequence	MDDWKPSPLIKPFGARKKRSWYLTWKYKLTNQRALRRFCQTGAVLFLLVT VIVNIKLILDTRRAISEANEDPEPEQDYDEALGRLEPPRRRGSGPRRVLD VEVYSSRSKVYVAVDGTTVLEDEAREQGRGIHVIVLNQATGHVMAKRVFD TYSPHEDEAMVLFLNMVAPGRVLICTVKDEGSFHLKDTAKALLRSLGSQA GPALGWRDTWAFVGRKGGPVFGEKHSKSPALSSWGDPVLLKTDVPLSSAE EAECHWADTELNRRRRRFCSKVEGYGSVCSCKDPTPIEFSPDPLPDNKVL NVPVAVIAGNRPNYLYRMLRSLLSAQGVSPQMITVFIDGYYEEPMDVVAL FGLRGIQHTPISIKNARVSQHYKASLTATFNLFPEAKFAVVLEEDLDIAV DFFSFLSQSIHLLEEDDSLYCISAWNDQGYEHTAEDPALLYRVETMPGLG WVLRRSLYKEELEPKWPTPEKLWDWDMWMRMPEQRRGRECIIPDVSRSYH FGIVGLNMNGYFHEAYFKKHKFNTVPGVQLRNVDSLKKEAYEVEVHRLLS EAEVLDHSKNPCEDSFLPDTEGHTYVAFIRMEKDDDFTTWTQLAKCLHIW DLDVRGNHRGLWRLFRKKNHFLMVGVPASPYSVKKPPSVTPIFLEPPPKE EGAPGAPEQT
Molecular weight	75 kDa
Amino acids	1 to 660

Specifications

Our Abpromise guarantee covers the use of **ab153784** 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
Endotoxin level	< 0.100 Eu/µg

Purity	>95% by SDS-PAGE .
Form	Liquid

Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.50 Constituents: 0.32% Tris HCl, 10% Glycerol, 0.88% Sodium chloride

General Info

Function	Participates in O-mannosyl glycosylation. May be responsible for the synthesis of the GlcNAc(beta1-2)Man(alpha1-)O-Ser/Thr moiety on alpha-dystroglycan and other O-mannosylated proteins. Is specific for alpha linked terminal mannose and does not have MGAT3, MGAT4, MGAT5, MGAT7 or MGAT8 activity.
Tissue specificity	Constitutively expressed. An additional weaker band is also detected in spinal cord, lymph node, and trachea. Expressed especially in astrocytes. Also expressed in immature and mature neurons.
Pathway	Protein modification; protein glycosylation.
Involvement in disease	Defects in POMGNT1 are the cause of muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies type A3 (MDDGA3) [MIM:253280]. MDDGA3 is an autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly and cerebellar hypoplasia. MDDGA3 patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Defects in POMGNT1 are the cause of muscular dystrophy-dystroglycanopathy congenital with mental retardation type B3 (MDDGB3) [MIM:613151]; also called muscular dystrophy congenital POMGNT1-related. MDDGB3 is an autosomal recessive disorder characterized by congenital muscular dystrophy associated with mental retardation and mild structural brain abnormalities. Clinical features include mental retardation, white matter changes, cerebellar cysts, pontine hypoplasia, myopia, optic atrophy, decreased alpha-dystroglycan on muscle biopsy and increased serum creatine kinase. Defects in POMGNT1 are the cause of muscular dystrophy-dystroglycanopathy limb-girdle type C3 (MDDGC3) [MIM:613157]; also called muscular dystrophy-dystroglycanopathy limb-girdle POMGNT1-related. MDDGC3 is a rare form of limb-girdle muscular dystrophy with normal cognition. Muscle biopsy shows dystrophic changes with variable staining for glycosylated alpha-dystroglycan.
Sequence similarities	Belongs to the glycosyltransferase 13 family.
Domain	Amino acid residues between 299-311 are important for both protein expression and enzymatic activity. The minimal catalytic domain is located between positions 299-651. Single amino acid substitutions in the stem domain from MEB patients abolished the activity of the membrane-bound form but not the soluble form. This suggests that the stem domain of the soluble form is unnecessary for activity, but that some amino acids play a crucial role in the membrane-bound form.
Cellular localization	Golgi apparatus membrane.

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