abcam

Product datasheet

Recombinant Human POMT1 protein ab161055

1 Image

Overview

Product name	Recombinant Human POMT1 protein
Protein length	Protein fragment

Description

Nature	Recombinant
Source	Wheat germ
Amino Acid Sequence	
Species	Human
Sequence	LPDWGYRQLEIVGEKLSRGYHGSTVWNVEEHRYGAS QEQRERERELHSPA QVDVSRNLSFMARFSELQWRMLALRSDDSEHKYSSS PLEWVTLDTNIA
Amino acids	483 to 580
Tags	proprietary tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab161055** in the following tested applications.

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

Applications	ELISA Western blot
Form	Liquid
Additional notes	Protein concentration is above or equal to 0.05 mg/ml.

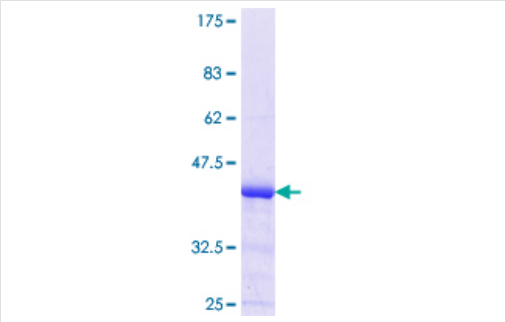
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Function	Transfers mannosyl residues to the hydroxyl group of serine or threonine residues. Coexpression of both POMT1 and POMT2 is necessary for enzyme activity, expression of either POMT1 or POMT2 alone is insufficient.
Tissue specificity	Widely expressed. Highly expressed in testis, heart and pancreas. Detected at lower levels in kidney, skeletal muscle, brain, placenta, lung and liver.
Pathway	Protein modification; protein glycosylation.
Involvement in disease	Defects in POMT1 are the cause of muscular dystrophy-dystroglycanopathy congenital with mental retardation type B1 (MDDGB1) [MIM:613155]; also called muscular dystrophy congenital POMT1-related. MDDGB1 is an autosomal recessive disorder characterized by congenital muscular dystrophy associated with mental retardation and mild structural brain abnormalities. Defects in POMT1 are the cause of muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies type A1 (MDDGA1) [MIM:236670]; also known as hydrocephalus-agyria-retinal dysplasia or HARD syndrome. MDDGA1 is an autosomal recessive disorder characterized by cobblestone lissencephaly, hydrocephalus, agyria, retinal displasia, with or without encephalocele. It is often associated with congenital muscular dystrophy and usually lethal within the first few months of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. Defects in POMT1 are the cause of muscular dystrophy-dystroglycanopathy limb-girdle type C1 (MDDGC1) [MIM:609308]; also called autosomal recessive limb-girdle muscular dystrophy with mental retardation. MDDGC1 is a novel form of recessive limb girdle muscular dystrophy with mild mental retardation without any obvious structural brain abnormality, associated with an abnormal alpha-dystroglycan pattern in the muscle. MDDGC1 is a significantly milder allelic form of WWS.
Sequence similarities	Belongs to the glycosyltransferase 39 family. Contains 3 MIR domains.
Cellular localization	Endoplasmic reticulum membrane.

Images



SDS-PAGE - Recombinant Human POMT1 protein (ab161055)

ab161055 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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