abcam

Product datasheet

Recombinant Human ALG12 protein ab163978

1 Image

Overview

Product name	Recombinant Human ALG12 protein
Protein length	Protein fragment

Description

Nature	Recombinant
Source	Wheat germ
Amino Acid Sequence	
Species	Human
Sequence	NYPGGVAMQRLHQLVPPQTDVLLHIDVAAAQTGVSRF LQVNSAWRYDKRE DVQPGTG
Amino acids	369 to 425
Tags	GST tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab163978** 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.

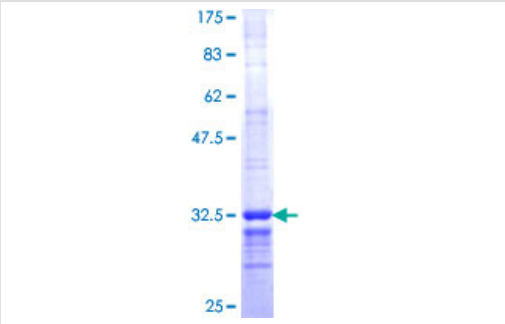
Preparation and Storage

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	Adds the eighth mannose residue in an alpha-1,6 linkage onto the dolichol-PP-oligosaccharide precursor (dolichol-PP-Man(7)GlcNAc(2)) required for protein glycosylation.
Tissue specificity	Expressed in fibroblasts.
Pathway	Protein modification; protein glycosylation.
Involvement in disease	Defects in ALG12 are the cause of congenital disorder of glycosylation type 1G (CDG1G) [MIM:607143]. CDGs are a family of severe inherited diseases caused by a defect in protein N-glycosylation. They are characterized by under-glycosylated serum proteins. These multisystem disorders present with a wide variety of clinical features, such as disorders of the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions.
Sequence similarities	Belongs to the glycosyltransferase 22 family.
Cellular localization	Endoplasmic reticulum membrane.

Images



SDS-PAGE - Recombinant Human ALG12 protein (ab163978)

ab163978 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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