abcam

Product datasheet

Recombinant Human FKRP protein ab153505

1 Image

Description

Product name	Recombinant Human FKRP protein
Expression system	Wheat germ
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	KAVEGDFFRVQYSESNHLHVDLWPFYPRNGVMTKDTWL DHRQDVEFPEHF LQPLVPLPFAGFVAQAPNNYRRFLELKFGPGVIENPQYPN PALLSLTGS
Amino acids	396 to 494
Tags	GST tag N-Terminus

Specifications

Our **Abpromise guarantee** covers the use of **ab153505** 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	

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

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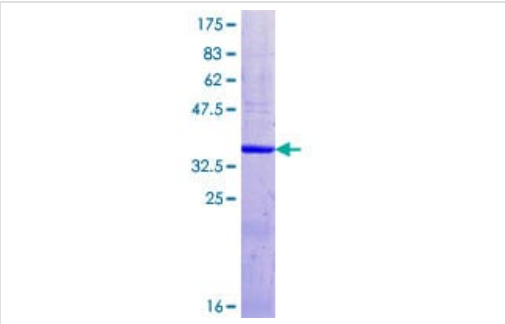
Function	Could be a transferase involved in the modification of glycan moieties of alpha-dystroglycan (DAG1).
Tissue specificity	Expressed predominantly in skeletal muscle, placenta, and heart and relatively weakly in brain, lung, liver kidney and pancreas.
Involvement in disease	Defects in FKRP are the cause of muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies type A5 (MDDGA5) [MIM:613153]. MDDGA5 is an autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. Defects in FKRP are the cause of muscular dystrophy-dystroglycanopathy congenital with or without mental retardation type B5 (MDDGB5) [MIM:606612]. MDDGB5 is a congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, a secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of MDDGB5 patients have brain involvements. Defects in FKRP are the cause of muscular dystrophy-dystroglycanopathy limb-girdle type C5 (MDDGC5) [MIM:607155]; also known as limb-girdle muscular dystrophy type 2I. MDDGC5 is an autosomal recessive disorder with age of onset ranging from childhood to adult life, and variable severity. Clinical features include proximal muscle weakness, waddling gait, calf hypertrophy, cardiomyopathy and respiratory insufficiency. A reduction of alpha-dystroglycan and laminin alpha-2 expression can be observed on skeletal muscle biopsy from MDDGC5 patients.
Sequence similarities	Belongs to the licD transferase family.
Post-translational modifications	N-glycosylated.
Cellular localization	Golgi apparatus. Secreted. Cell membrane > sarcolemma. Rough endoplasmic reticulum. According to some studies the N-terminal hydrophobic domain is cleaved after translocation to the Golgi apparatus and the protein is secreted. According to others the N-terminal hydrophobic domain is a transmembrane domain and the protein is a type II transmembrane type targeted to the Golgi apparatus by a non-cleavable signal anchor sequence. Localization at the cell membrane may require the presence of dystroglycan. At the Golgi apparatus localizes most likely at the cis-compartment. Detected in rough endoplasmic reticulum in myocytes. In general, mutants associated with severe clinical phenotypes are retained within the endoplasmic reticulum.

Images



SDS-PAGE - Recombinant Human FKRP protein (ab153505)

ab153505 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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