

FKRP Rabbit pAb

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Product Information

Application WB, IHC-P, IHC-F, IF

Primary Accession 09H9S5

Reactivity Pig, Mouse, Dog

Host Rabbit Clonality Polyclonal Calculated MW 54568 **Physical State** Liquid

Immunogen KLH conjugated synthetic peptide derived from human FKRP

Epitope Specificity 1-100/495

affinity purified by Protein A **Purity**

Buffer SUBCELLULAR LOCATION 0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol. 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.

SIMILARITY Post-translational modifications **DISEASE**

Belongs to the licD transferase family.

N-glycosylated.

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.

Important Note This product as supplied is intended for research use only, not for use in

human, therapeutic or diagnostic applications.

Background Descriptions This gene encodes a protein which is targeted to the medial Golgi apparatus

and is necessary for posttranslational modification of dystroglycan. Mutations in this gene have been associated with congenital muscular dystrophy, mental retardation, and cerebellar cysts. Several alternatively spliced transcript variants of this gene have been described, but the full-length nature of some of these variants has not been determined. [provided by RefSeq, Oct 2008]

Additional Information

Gene ID 79147

Other Names Ribitol 5-phosphate transferase FKRP, 2.7.8.-, Fukutin-related protein,

Ribitol-5-phosphate transferase, FKRP (HGNC:17997)

Target/Specificity Expressed predominantly in skeletal muscle, placenta, and heart and

relatively weakly in brain, lung, liver kidney and pancreas.

Dilution WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500

Storage Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When

reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody

is stable for at least two weeks at 2-4 °C.

Protein Information

Name FKRP (HGNC:17997)

Function Catalyzes the transfer of a ribitol 5-phosphate from CDP-L- ribitol to the

ribitol 5-phosphate previously attached by FKTN/fukutin to the phosphorylated O-mannosyl trisaccharide (N-acetylgalactosamine-

beta-3-N-acetylglucosamine-beta-4-(phosphate-6-)mannose), a carbohydrate

structure present in alpha-dystroglycan (DAG1) (PubMed:26923585,

PubMed:<u>27194101</u>, PubMed:<u>29477842</u>, PubMed:<u>31949166</u>). This constitutes the second step in the formation of the ribose 5- phosphate tandem repeat which links the phosphorylated O-mannosyl trisaccharide to the ligand binding moiety composed of repeats of 3- xylosyl-alpha-1,3-glucuronic acid-beta-1 (PubMed:<u>25279699</u>, PubMed:<u>26923585</u>, PubMed:<u>27194101</u>,

PubMed:29477842, PubMed:31949166).

Cellular Location Golgi apparatus membrane; Single-pass type II membrane protein. Secreted.

Cell membrane, sarcolemma {ECO:0000250 | UniProtKB:Q8CG64}. Rough endoplasmic reticulum. Cytoplasm {ECO:0000250 | UniProtKB:Q8CG64}. Note=According to some studies the N- terminal hydrophobic domain is cleaved after translocation to the Golgi apparatus and the protein is secreted (PubMed:19900540). Localization at the cell membrane may require the presence of dystroglycan (By similarity). At the Golgi apparatus localizes to the middle-to-trans- cisternae, as assessed by MG160 colocalization. Detected in

rough endoplasmic reticulum in myocytes (PubMed:17554798,

PubMed:21886772) In general, mutants associated with severe clinical

phenotypes are retained within the endoplasmic reticulum (PubMed:15213246) {ECO:0000250 | UniProtKB:Q8CG64,

ECO:0000269 | PubMed:15213246, ECO:0000269 | PubMed:17554798, ECO:0000269 | PubMed:19900540, ECO:0000269 | PubMed:21886772}

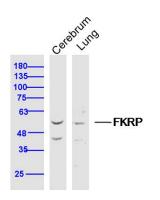
Tissue Location

Expressed in the retina (at protein level) (PubMed:29416295). Expressed predominantly in skeletal muscle, placenta, and heart and relatively weakly in brain, lung, liver, kidney, and pancreas (PubMed:11592034).

Background

This gene encodes a protein which is targeted to the medial Golgi apparatus and is necessary for posttranslational modification of dystroglycan. Mutations in this gene have been associated with congenital muscular dystrophy, mental retardation, and cerebellar cysts. Several alternatively spliced transcript variants of this gene have been described, but the full-length nature of some of these variants has not been determined. [provided by RefSeq, Oct 2008]

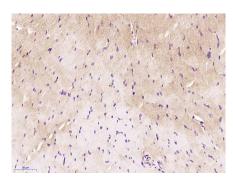
Images



Sample:

Cerebrum (Mouse) Lysate at 40 ug Lung (Mouse) Lysate at 40 ug Primary: Anti-FKRP (AP56118) at 1/300 dilution Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution

Predicted band size: 55 kD Observed band size: 55 kD



Paraformaldehyde-fixed, paraffin embedded (Rat skeletal muscle); Antigen retrieval by boiling in sodium citrate buffer (pH6.0) for 15min; Block endogenous peroxidase by 3% hydrogen peroxide for 20 minutes; Blocking buffer (normal goat serum) at 37°C for 30min; Antibody incubation with (FKRP) Polyclonal Antibody, Unconjugated (AP56118) at 1:400 overnight at 4°C, followed by operating according to SP Kit(Rabbit) (sp-0023) instructions and DAB staining.

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.