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FKRP Rabbit pAb

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Catalog # AP56118

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	Q9H9S5
Reactivity	Mouse, Rat
Predicted	Human, Dog, Pig, Rabbit
Host	Rabbit
Clonality	Polyclonal
Calculated MW	54568
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human FKRP
Epitope Specificity	1-100/495
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	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	Belongs to the licD transferase family.
Post-translational modifications	N-glycosylated.
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.
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,IF=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:27194101, PubMed:29477842, PubMed:31949166). 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:25279699, PubMed:26923585, PubMed:27194101, 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}

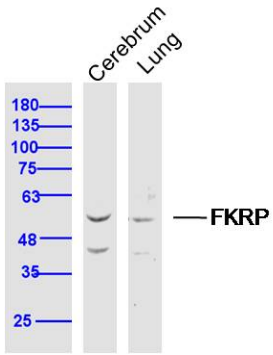
Tissue Location

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).

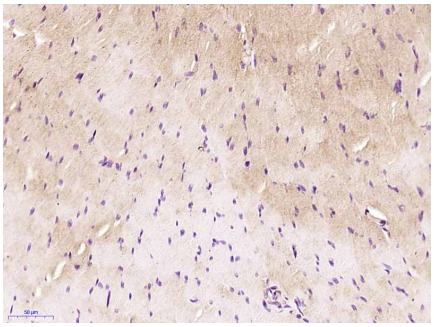
Background

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) instructionsand DAB staining.