Product Name: FKRP Rabbit Polyclonal Antibody
Catalog #: APRab11015

Summary

Production Name	FKRP Rabbit Polyclonal Antibody
Description	Rabbit Polyclonal Antibody
Host	Rabbit
Application	WB
Reactivity	Human,Mouse,Rat

Performance

Conjugation	Unconjugated
Modification	Unmodified
Isotype	IgG
Clonality	Polyclonal
Form	Liquid
Storage	Store at 4°C short term. Aliquot and store at -20°C long term. Avoid freeze/thaw cycles.
Buffer	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Purification	Affinity purification

Immunogen

Gene Name	FKRP
Alternative Names	FKRP; Fukutin-related protein
Gene ID	79147.0
SwissProt ID	Q9H9S5.The antiserum was produced against synthesized peptide derived from human FKRP. AA range:1-50

Application

Dilution Ratio	WB 1:500-1:2000. ELISA: 1:20000.
Molecular Weight	50kD

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],disease:Defects in FKRP are the cause of congenital muscular dystrophy type 1C (MDC1C) [MIM:606612]. Congenital muscular dystrophies (CMD) are a heterogeneous group of autosomal recessive disorders characterized by hypotonia, muscle weakness, and joint contractures that present at birth or during the first 6 months of life and have dystrophic changes on skeletal muscle biopsy. Mental retardation with or without structural CNS changes may accompany some forms. MDC1C is a form of CMD with onset in the first weeks of life and 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 MDC1C patients have brain involvements.,disease:Defects in FKRP are the cause of limb-girdle muscular dystrophy type 2I (LGMD2I) [MIM:607155]. LGMD2I 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 LGMD2I patients.,disease:Defects in FKRP may be a cause of muscle-eye-brain disease (MEB) [MIM:253280]. MEB is an autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly and cerebellar hypoplasia. MEB patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks.,disease:Defects in FKRP may be a cause of Walker-Warburg syndrome (WWS) [MIM:236670]; also known as hydrocephalus-agyria-retinal dysplasia or HARD syndrome. WWS 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.,function:Could be a transferase involved in the modification of glycan moieties of alpha-dystroglycan (DAG1).,online information:GlycoGene database,similarity:Belongs to the licD transferase family.,tissue specificity:Expressed predominantly in skeletal muscle, placenta, and heart and relatively weakly in brain, lung, liver kidney and pancreas.,

Research Area

Image Data

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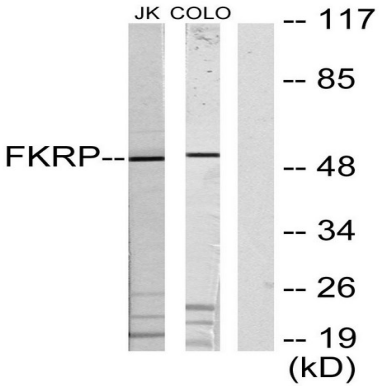

Western blot analysis of lysates from Jurkat and COLO205 cells, using FKRP Antibody. The lane on the right is blocked with the synthesized peptide.

Note

For research use only.