FKRP Rabbit pAb

CatalogNo: YT1715

Key Features

Host Species
- Rabbit

Reactivity
- Human,Mouse,Rat

Applications
- WB,IF,ELISA

MW
- 50kD (Observed)

Isotype
- IgG

Recommended Dilution Ratios

WB 1:500-1:2000
IF 1:200-1:1000
ELISA 1:20000
Not yet tested in other applications.

Storage

Storage*	-15°C to -25°C/1 year(Do not lower than -25°C)
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.

Basic Information

Clonality	Polyclonal

Immunogen Information

Immunogen	The antiserum was produced against synthesized peptide derived from human FKRP. AA range:1-50
Specificity	FKRP Polyclonal Antibody detects endogenous levels of FKRP protein.

Target Information

Gene name FKRP

Protein Name Fukutin-related protein

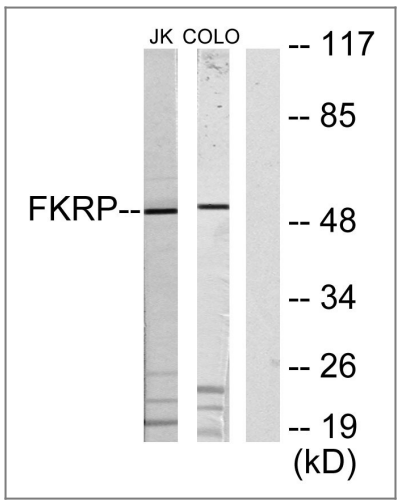
Organism	Gene ID	UniProt ID
Human	79147;	Q9H9S5;
Mouse	243853;	Q8CG64;

Cellular Localization Golgi apparatus membrane ; Single-pass type II membrane protein . Secreted . Cell membrane, sarcolemma . Rough endoplasmic reticulum . Cytoplasm . 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). .

Tissue specificity 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).

Function 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.,

Validation Data



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

Contact information

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Please scan the QR code to access additional product information:
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