产品名称：FKRP Rabbit Polyclonal Antibody
产品货号：APRab11015

产品概述（Summary）

产品名称（Production Name）	FKRP Rabbit Polyclonal Antibody
描述（Description）	Rabbit polyclonal Antibody
宿主（Host）	Rabbit
应用（Application）	WB,ICC/IF,ELISA
种属反应性（Reactivity）	Human,Mouse,Rat

产品性能（Performance）

偶联物（Conjugation）	Unconjugated
修饰（Modification）	Unmodified
同种型（Isotype）	IgG
克隆（Clonality）	Polyclonal
形式（Form）	Liquid
存放说明（Storage）	Store at 4°C short term. Aliquot and store at -20°C long term. Avoid freeze/thaw cycles.
储存溶液（Buffer）	Liquid in PBS containing 50% glycerol, 0.5% protective protein and 0.02% New type preservative N.
纯化方式（Purification）	Affinity purification

免疫原信息（Immunogen）

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

产品应用（Application）

稀释比（Dilution Ratio）	WB 1:500-1:2000,ICC/IF 1:200-1:1000,ELISA 1:10000-1:20000
蛋白分子量（Molecular Weight）	50kDa

研究背景（Background）

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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.,

研究领域（Research Area）

图片（Image Data）

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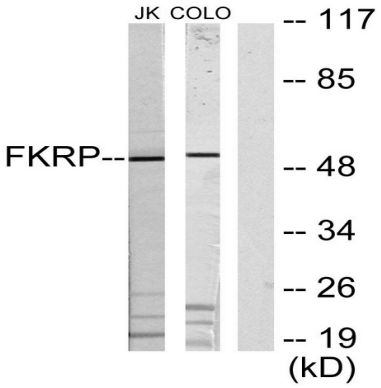

Western blot analysis of lysates from Jurkat and COLO205 cells, using FKRP Antibody. The lane on the right is blocked with the synthesized peptide.

注意事项（Note）

For research use only .