



FKRP Monoclonal Antibody

Catalog No	YP-mAb-01712
Isotype	IgG
Reactivity	Human;Mouse;Rat
Applications	WB
Gene Name	FKRP
Immunogen	The antiserum was produced against synthesized peptide derived from human FKRP. AA range:1-50
Specificity	FKRP Monoclonal Antibody detects endogenous levels of FKRP protein.
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.
Source	Monoclonal, Mouse,IgG
Purification	The antibody was affinity-purified from mouse antiserum by affinity-chromatography using epitope-specific immunogen.
Dilution	WB 1:500-1:2000
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	FKRP;Fukutin-related protein
Observed Band	50kD
Calculated Molecular Weight	50kD
Cell Pathway	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

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],

matters needing attention

Avoid repeated freezing and thawing!

Usage suggestions

This product can be used in immunological reaction related experiments. For more information, please consult technical personnel.

Products Images

