FKRP Antibody – Rabbit Polyclonal

PRODUCT OVERVIEW

Product Name	FKRP, Polyclonal Antibody
Full Product Name	FKRP, ID (FKRP, Fukutin-related protein) (MaxLight 490)
Product Gene Name	anti-FKRP antibody
Product Synonym Names	FKRP; Fukutin-related protein
Clonality	Polyclonal
Isotype	IgG
Host	Rabbit
Species Reactivity	Human
Form/Format	Supplied as a liquid in PBS, pH 7.2. No preservative added. Labeled with MaxLight490.
Purity/Purification	Purified by Protein A Affinity Chromatography.
Preparation and Storage	Store product at 4 degree C in the dark. DO NOT FREEZE! Stable at 4 degree C for 12 months after receipt as an undiluted liquid. Dilute required amount only prior to immediate use. Further dilutions can be made in assay buffer. Caution: MaxLight490 conjugates are sensitive to light. For maximum recovery of product, centrifuge the original vial prior to removing the cap.
Molecular Weight	54,568 Da
OMIM	606596
3D Structure	ModBase 3D Structure for Q9H9S5
Immunogen	FKRP antibody is generated from rabbits immunized with a KLH conjugated synthetic peptide between 236-265 amino acids from the Central region of human FKRP.
Conjugate	MaxLight490
Note	Preservative Free
Product Categories/Family for anti-FKRP antibody	Antibodies; Proteins
Applications Tested/Suitable for anti-FKRP antibody	Western Blot (WB), FLISA
Application Notes for anti-FKRP antibody	WB: 1:100-500 FLISA: 1:1,000 Applications are based on unconjugated antibody.
Related Product Information for anti-FKRP antibody	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].
Product References and Citations for anti-FKRP antibody	Kawahara, G., et al. Hum. Mol. Genet. 19(4):623-633(2010). Crowther-Swanepoel, D., et al. Nat. Genet. 42(2):132-136(2010). Lu, P.J., et al. Biochim. Biophys. Acta 1802(2):253-258(2010). Hanisch, F., et al. J. Neurol. 257(2):300-301(2010). Bourteel, H., et al. J. Neurol. Neurosurg. Psychiatr. 80(12):1405-1408(2009).
Other Notes	Small volumes of anti-FKRP antibody vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply.

Searchable Terms for FKRP purchase	MBS6299391 is an antibody targeting FKRP. Also known as FKRP, ID (FKRP, Fukutin-related protein) (MaxLight 490). Reactive in Human. Host: Rabbit.

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for FKRP. It may not necessarily be applicable to this product.

NCBI GI #	13236528
NCBI GeneID	79147
NCBI Accession #	NP_077277.1
NCBI Related Accession #	NP_001034974.1
UniProt Primary Accession #	Q9H9S5
UniProt Secondary Accession #	A8K5G7
UniProt Related Accession #	Q9H9S5
GenBank Nucleotide #	NM_024301.4
NCBI Official Full Name	fukutin-related protein
NCBI Official Synonym Full Names	fukutin related protein
NCBI Official Symbol	FKRP
NCBI Official Synonym Symbols	MDC1C; LGMD2I; MDDGA5; MDDGB5; MDDGC5
UniProt Protein Name	Fukutin-related protein
Protein Family	Fukutin-related protein
UniProt Gene Name	FKRP
UniProt Entry Name	FKRP_HUMAN
NCBI Protein Information	fukutin-related protein
NCBI Summary for FKRP	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]

UniProt Comments for FKRP	FKRP: Could be a transferase involved in the modification of glycan moieties of alpha-dystroglycan (DAG1). Defects in FKRP are the cause of muscular dystrophy- dystroglycanopathy congenital with brain and eye anomalies type A5 (MDDGA5). 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). 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); 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. Belongs to the LicD transferase family. Protein type: Membrane protein, integral; Transferase; EC 2.-.-.- Chromosomal Location of Human Ortholog: 19q13.32 Cellular Component: Golgi membrane; dystrophin-associated glycoprotein complex; Golgi apparatus; extracellular space; rough endoplasmic reticulum; integral to membrane; sarcolemma Molecular Function: transferase activity Biological Process: protein amino acid O-linked mannosylation; protein processing Disease: Muscular Dystrophy-dystroglycanopathy (congenital With Or Without Mental Retardation), Type B, 5; Muscular Dystrophy-dystroglycanopathy (congenital With Brain And Eye Anomalies), Type A, 5; Muscular Dystrophy-dystroglycanopathy (limb-girdle), Type C, 5; Muscular Dystrophy-dystroglycanopathy (congenital With Brain And Eye Anomalies), Type A, 1
Research Articles on FKRP	**1.** This study demonstrated that the higher frequency of LGMD2I with cardiomyopathy in mutation of FKRP in Taiwanese patients.
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