FKTN Peptide - N-Terminal Region Blocking Peptide

PRODUCT OVERVIEW

Product Name	FKTN, Blocking Peptide
Full Product Name	FKTN Peptide - N-terminal region
Product Gene Name	FKTN blocking peptide
Product Synonym Gene Name	CMD1X; FCMD; LGMD2M; MDDGA4; MDDGB4; MDDGC4
Sequence	Synthetic peptide located within the following region: LLTLTSSAFL LFQLYYYKHY LSTKNGAGLS KSKGSRIGFD STQWRAVKKF
Species Reactivity	Human
Form/Format	Lyophilized powder
Preparation and Storage	Add 100ul of sterile PBS. Final peptide concentration is 1 mg/ml in PBS. For longer periods of storage, store at -20 degree C. Avoid repeat freeze-thaw cycles.
Antibody/Peptide Pairs	FKTN peptide (MBS3232597) is used for blocking the activity of FKTN antibody (MBS3207632)
Molecular Weight	47kDa
OMIM	phenotype 613152
Product Categories/Family for FKTN blocking peptide	Peptide
Applications Tested/Suitable for FKTN blocking peptide	Western Blot (WB)
Related Product Information for FKTN blocking peptide	This is a synthetic peptide designed for use in combination with anti-FKTN Antibody, made Target Description: FKTN regulates the migration and assembly of neurons during cortical histogenesis. Fukuyama congenital muscular dystrophy results from mutations in its gene.
Other Notes	Small volumes of FKTN blocking peptide 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 FKTN purchase	MBS3232597 is a blocking peptide targeting FKTN. Also known as FKTN Peptide - N-terminal region. Reactive in Human.

ADDITIONAL DETAILS

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

NCBI GI #	119395714
NCBI GeneID	2218
NCBI Accession #	NP_006722
UniProt Primary Accession #	O75072
UniProt Related Accession #	O75072
GenBank Nucleotide #	NM_006731
NCBI Official Full Name	fukutin isoform a

NCBI Official Synonym Full Names	fukutin
NCBI Official Symbol	FKTN
NCBI Official Synonym Symbols	FCMD; CMD1X; LGMD2M; MDDGA4; MDDGB4; MDDGC4; LGMDR13
UniProt Protein Name	Fukutin
UniProt Synonym Protein Names	Fukuyama-type congenital muscular dystrophy protein
Protein Family	Fukutin
UniProt Gene Name	FKTN
UniProt Synonym Gene Names	FCMD
UniProt Entry Name	FKTN_HUMAN
NCBI Protein Information	fukutin
NCBI Summary for FKTN	The protein encoded by this gene is a putative transmembrane protein that is localized to the cis-Golgi compartment, where it may be involved in the glycosylation of alpha-dystroglycan in skeletal muscle. The encoded protein is thought to be a glycosyltransferase and could play a role in brain development. Defects in this gene are a cause of Fukuyama-type congenital muscular dystrophy (FCMD), Walker-Warburg syndrome (WWS), limb-girdle muscular dystrophy type 2M (LGMD2M), and dilated cardiomyopathy type 1X (CMD1X). Alternatively spliced transcript variants have been found for this gene. [provided by RefSeq, Nov 2010]
UniProt Comments for FKTN	FCMD: May be a glycosyltransferase which participates in glycosylation of alpha-dystroglycan/DAG1. May interact with and reinforce a large complex encompassing the outside and inside of muscle membranes. Could be involved in brain development. Defects in FKTN are the cause of muscular dystrophy- dystroglycanopathy congenital with brain and eye anomalies type A4 (MDDGA4); also called congenital muscular dystrophy Fukuyama type (FCMD) or Walker-Warburg syndrome FKTN-related. MDDGA4 is an autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies. Patients suffer from generalized skeletal muscle weakness and hypotonia from early infancy, mental retardation and seizures. Occasional features include optic atrophy, retinal detachment, cardiomyopathy. Defects in FKTN are the cause of muscular dystrophy- dystroglycanopathy congenital without mental retardation type B4 (MDDGB4). An autosomal recessive disorder characterized by congenital muscular dystrophy and evidence of dystroglycanopathy. Features included increased serum creatine kinase, generalized weakness, mild white matter changes on brain MRI in some cases, and absence of mental retardation. Defects in FKTN are the cause of muscular dystrophy-dystroglycanopathy limb-girdle type C4 (MDDGC4). MDDGC4 is an autosomal recessive degenerative myopathy characterized by progressive weakness of the pelvic and shoulder girdle muscles and elevated serum creatine kinase. The severity of the disease depends on age at onset which may vary from early to late childhood or even adulthood. MDDGC4 is a novel form of LGMD2 and has no brain involvement and a remarkable clinical response to corticosteroids. Defects in FKTN are the cause of cardiomyopathy dilated type 1X (CMD1X); also called dilated cardiomyopathy with mild or no proximal muscle weakness. Dilated cardiomyopathy is a disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. Belongs to the LicD transferase family. Protein type: EC 2.-.-.-; Transferase; Membrane protein, integral; Endoplasmic reticulum Chromosomal Location of Human Ortholog: 9q31.2 Cellular Component: Golgi membrane; Golgi apparatus; extracellular space; cis-Golgi network; endoplasmic reticulum; integral to membrane; nucleus Molecular Function: transferase activity Biological Process: negative regulation of cell proliferation; nervous system development; negative regulation of JNK cascade; muscle development; protein amino acid O-linked mannosylation; regulation of protein amino acid glycosylation Disease: Cardiomyopathy, Dilated, 1x; Muscular Dystrophy-dystroglycanopathy (congenital With Brain And Eye Anomalies), Type A, 4; Muscular Dystrophy-dystroglycanopathy (limb-girdle), Type C, 4; Muscular Dystrophy-dystroglycanopathy (congenital Without Mental Retardation), Type B, 4; Muscular Dystrophy-dystroglycanopathy (congenital With Brain And Eye Anomalies), Type A, 1

Research Articles on FKTN	**1.** The results suggest that fukutin and FKRP not only participate in the synthesis of O-mannosyl glycans added to alpha-dystroglycan in the endoplasmic reticulum and Golgi complex, but that they could also play a role, that remains to be established, in the nucleus of retinal neurons.
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