

FKTN Antibody (Center) Blocking peptide

Synthetic peptide Catalog # BP12786c

Specification

FKTN Antibody (Center) Blocking peptide - Product Information

Primary Accession

FKTN Antibody (Center) Blocking peptide - Additional Information

Gene ID 2218

Other Names

Fukutin, 2---, Fukuyama-type congenital muscular dystrophy protein, FKTN, FCMD

Format

Peptides are lyophilized in a solid powder format. Peptides can be reconstituted in solution using the appropriate buffer as needed.

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Storage

Maintain refrigerated at 2-8°C for up to 6 months. For long term storage store at -20°C.

Precautions

This product is for research use only. Not for use in diagnostic or therapeutic procedures.

FKTN Antibody (Center) Blocking peptide - Protein Information

Name FKTN (HGNC:3622)

Function

Catalyzes the transfer of a ribitol-phosphate from CDP- ribitol to the distal N-acetylgalactosamine of the phosphorylated O- mannosyl trisaccharide (N-acetylgalactosamine-beta-3-Nacetylglucosamine-beta-4-(phosphate-6-)mannose), a carbohydrate structure present in alpha-dystroglycan (DAG1) (PubMed:26923585, PubMed:29477842, PubMed:27194101). This constitutes the first step in the formation of the ribitol 5-phosphate tandem repeat which links the phosphorylated O-mannosyl trisaccharide to the ligand binding moiety composed of repeats of 3-xylosyl-alpha-1,3-glucuronic acid-beta-1 (PubMed: 17034757, PubMed:25279699, PubMed:26923585, PubMed:29477842, PubMed:27194101). Required for normal location of POMGNT1 in Golgi membranes, and for normal POMGNT1 activity (PubMed: 17034757). May interact with and reinforce a large complex encompassing the outside and inside of muscle membranes (PubMed:25279699).



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Could be involved in brain development (Probable).

Cellular Location

Golgi apparatus membrane; Single-pass type II membrane protein. Cytoplasm {ECO:0000250|UniProtKB:Q8R507}. Nucleus {ECO:0000250|UniProtKB:Q8R507}. Note=In retinal tissue, does not localize with the Golgi apparatus. {ECO:0000250|UniProtKB:Q8R507}

Tissue Location

Expressed in the retina (at protein level) (PubMed:29416295). Widely expressed with highest expression in brain, heart, pancreas and skeletal muscle (PubMed:11115853). Expressed at similar levels in control fetal and adult brain (PubMed:11115853) Expressed in migrating neurons, including Cajar-Retzius cells and adult cortical neurons, as well as hippocampal pyramidal cells and cerebellar Purkinje cells (PubMed:11115853). No expression observed in the glia limitans, the subpial astrocytes (which contribute to basement membrane formation) or other glial cells (PubMed:11115853)

FKTN Antibody (Center) Blocking peptide - Protocols

Provided below are standard protocols that you may find useful for product applications.

• Blocking Peptides

FKTN Antibody (Center) Blocking peptide - Images

FKTN Antibody (Center) Blocking peptide - Background

The protein encoded by this gene is a putative transmembrane protein that is localized to the cis-Golgicompartment, where it may be involved in the glycosylation ofalpha-dystroglycan in skeletal muscle. The encoded protein isthought to be a glycosyltransferase and could play a role in braindevelopment. Defects in this gene are a cause of Fukuyama-typecongenital muscular dystrophy (FCMD), Walker-Warburg syndrome(WWS), limb-girdle muscular dystrophy type 2M (LGMD2M), and dilatedcardiomyopathy type 1X (CMD1X). Alternatively spliced transcriptvariants have been found for this gene.

FKTN Antibody (Center) Blocking peptide - References

Lim, B.C., et al. Neuromuscul. Disord. 20(8):524-530(2010)Saredi, S., et al. Muscle Nerve 39(6):845-848(2009)Chang, W., et al. Prenat. Diagn. 29(6):560-569(2009)Mercuri, E., et al. Neurology 72(21):1802-1809(2009)Puckett, R.L., et al. Neuromuscul. Disord. 19(5):352-356(2009)