10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

FKTN Antibody (Center)

Affinity Purified Rabbit Polyclonal Antibody (Pab)
Catalog # AP12786c

Product Information

Application	IHC-P, WB, E
Primary Accession	O75072
Other Accession	Q60HG0, NP_001073270.1, NP_006722.2
Reactivity	Human
Predicted	Monkey
Host	Rabbit
Clonality	Polyclonal
Isotype	Rabbit IgG
Clone Names	RB31283
Calculated MW	53724
Antigen Region	177-206

Additional Information

Gene ID	2218
Other Names	Fukutin, 2---, Fukuyama-type congenital muscular dystrophy protein, FKTN, FCMD
Target/Specificity	This FKTN antibody is generated from rabbits immunized with a KLH conjugated synthetic peptide between 177-206 amino acids from the Central region of human FKTN.
Dilution	IHC-P~~1:100~500 WB~~1:1000 E~~Use at an assay dependent concentration.
Format	Purified polyclonal antibody supplied in PBS with 0.09% (W/V) sodium azide. This antibody is purified through a protein A column, followed by peptide affinity purification.
Storage	Maintain refrigerated at 2-8°C for up to 2 weeks. For long term storage store at -20°C in small aliquots to prevent freeze-thaw cycles.
Precautions	FKTN Antibody (Center) is for research use only and not for use in diagnostic or therapeutic procedures.

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-N-acetylglucosamine-beta-4-(phosphate-6-)mannose), a carbohydrate structure present in alpha-dystroglycan (DAG1) (PubMed:26923585, PubMed:27194101, PubMed:29477842). 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:27194101, PubMed:29477842). 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). 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)

Background

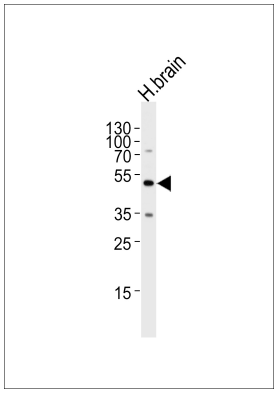
The protein encoded by this gene is a putative transmembrane protein that is localized to the cis-Golgi compartment, where it may be involved in the glycosylation of alpha-dystroglycan in skeletal muscle. The encoded protein is thought to be a glycosyltransferase and could play a role in brain development. 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.

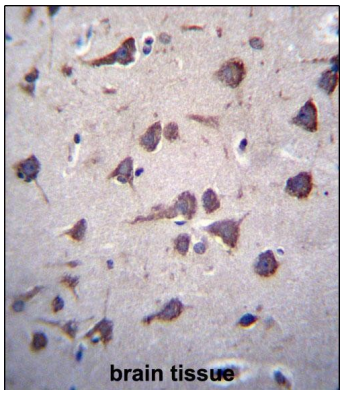
References

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)

Images

Western blot analysis of lysate from human brain tissue lysate, using FKTN Antibody (Center)(Cat. #AP12786c). AP12786c was diluted at 1:1000 at each lane. A goat anti-rabbit IgG H&L(HRP) at 1:5000 dilution was used as the secondary antibody. Lysate at 35ug per lane.





FKTN Antibody (Center) (Cat. #AP12786c)immunohistochemistry analysis in formalin fixed and paraffin embedded human brain tissue followed by peroxidase conjugation of the secondary antibody and DAB staining.This data demonstrates the use of FKTN Antibody (Center) for immunohistochemistry. Clinical relevance has not been evaluated.