FKTN Polyclonal Antibody

Catalog No	BYab-07313
Isotype	IgG
Reactivity	Human;Mouse
Applications	WB;ELISA
Gene Name	FKTN FCMD
Protein Name	Fukutin (EC 2.-.-.-) (Fukuyama-type congenital muscular dystrophy protein)
Immunogen	Synthesized peptide derived from human protein . at AA range: 111-160
Specificity	FKTN Polyclonal Antibody detects endogenous levels of protein.
Formulation	Liquid in PBS containing 50% glycerol, and 0.02% sodium azide.
Source	Polyclonal, Rabbit,IgG
Purification	The antibody was affinity-purified from rabbit antiserum by affinity-chromatography using epitope-specific immunogen.
Dilution	WB 1:500-2000 ELISA 1:5000-20000
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	
Observed Band	50kD
Cell Pathway	Golgi apparatus membrane ; Single-pass type II membrane protein . Cytoplasm . Nucleus . In retinal tissue, does not localize with the Golgi apparatus. .
Tissue Specificity	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).
Function	disease:Defects in FKTN are a cause of Walker-Warburg syndrome (WWS) [MIM:236670]; also known as hydrocephalus-agyria-retinal dysplasia or HARD syndrome. WWS is an autosomal recessive disorder characterized by cobblestone lissencephaly, hydrocephalus, agyria, retinal displasia, with or without encephalocele. It is often associated with congenital muscular dystrophy

	and usually lethal within the first few months of life.,disease:Defects in FKTN are the cause of cardiomyopathy dilated type 1X (CMD1X) [MIM:611615]; 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.,disease:Defects in FKTN are the cause of congenital muscular dystrophy Fukuyama type (FCMD) [MIM:253800
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. [provided by RefSeq, Nov 2010],
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.

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