



# FKTN Monoclonal Antibody

<b>Catalog No</b>	BYmab-07313
<b>Isotype</b>	IgG
<b>Reactivity</b>	Human;Mouse
<b>Applications</b>	WB
<b>Gene Name</b>	FKTN FCMD
<b>Protein Name</b>	Fukutin (EC 2.-.-.) (Fukuyama-type congenital muscular dystrophy protein)
<b>Immunogen</b>	Synthesized peptide derived from human protein . at AA range: 111-160
<b>Specificity</b>	FKTN Monoclonal Antibody detects endogenous levels of protein.
<b>Formulation</b>	Liquid in PBS containing 50% glycerol, and 0.02% sodium azide.
<b>Source</b>	Monoclonal, Mouse,IgG
<b>Purification</b>	The antibody was affinity-purified from mouse antiserum by affinity-chromatography using epitope-specific immunogen.
<b>Dilution</b>	WB 1:500-2000
<b>Concentration</b>	1 mg/ml
<b>Purity</b>	≥90%
<b>Storage Stability</b>	-20°C/1 year
<b>Synonyms</b>	
<b>Observed Band</b>	50kD
<b>Cell Pathway</b>	Golgi apparatus membrane ; Single-pass type II membrane protein . Cytoplasm . Nucleus . In retinal tissue, does not localize with the Golgi apparatus. .
<b>Tissue Specificity</b>	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 Cajal-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).
<b>Function</b>	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 dysplasia, with or without encephalocele. It is often associated with congenital muscular dystrophy

Nanjing BYabscience technology Co.,Ltd



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.

## Products Images

Nanjing BYabscience technology Co.,Ltd