Dysferlin 蛋白抗体

产品货号： mIR2429

英文名称： Dysferlin

中文名称： Dysferlin 蛋白抗体

别　　名： DMAT; DYSF; Dysferlin (Dystrophy associated fer 1 like protein) (Fer 1 like protein 1); Dysferlin limb girdle muscular dystrophy 2B (autosomal recessive); Dysferlin limb girdle muscular dystrophy 2B; Dystrophy associated fer 1 like 1; Dystrophy associated fer 1 like protein; Dystrophy associated fer1 like 1; Dystrophy associated fer1 like protein; Fer 1 like protein 1; Fer1 like protein 1; FER1L1; FLJ00175; FLJ90168; LGMD 2B; LGMD2B; Limb girdle muscular dystrophy 2B (autosomal recessive) ; Limb girdle muscular dystrophy 2B; Miyoshi myopathy; MM; DYSF_HUMAN.

研究领域： 免疫学　通道蛋白

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Dog, Pig, Cow, Horse, Rabbit, Guinea Pig,

产品应用： ELISA=1:500-1000 Flow-Cyt=3ug/Test
not yet tested in other applications.
optimal dilutions/concentrations should be determined by the end user.

分 子 量： **237kDa**

细胞定位： 细胞膜

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human Dysferlin:1901-2119/2119

亚　　型： IgG

纯化方法： affinity purified by Protein A

储 存 液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： Store at -20 ° C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20° C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 ° C.

PubMed： PubMed

产品介绍： The protein encoded by this gene belongs to the ferlin family and is a skeletal muscle protein found associated with the sarcolemma. It is involved in muscle contraction and contains C2 domains that play a role in calcium-mediated membrane fusion events, suggesting that it may be involved in membrane regeneration and repair. In addition, the protein encoded by this gene binds caveolin-3, a skeletal muscle membrane protein which is important in the formation of caveolae. Specific mutations in this gene have been shown to cause autosomal recessive limb girdle muscular dystrophy type 2B (LGMD2B) as well as Miyoshi myopathy. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Aug 2008].

Function:

Key calcium ion sensor involved in the Ca(2+)-triggered synaptic vesicle-plasma membrane fusion. Plays a role in the sarcolemma repair mechanism of both skeletal muscle and cardiomyocytes that permits rapid resealing of membranes disrupted by mechanical stress (By similarity).

Subunit:

Interacts with CACNA1S. Interacts with ANXA1; the interaction is Ca(2+)- and injury state-dependent. Interacts with ANXA2; the interaction is Ca(2+)- and injury state-dependent. Interacts with CACNA1S and PARVB. Interacts with TRIM72/MG53; interaction is required for transport to sites of cell injury during repair patch formation (By

similarity). Interacts with CAV3 and PARVB. Interacts with AHNAK; the interaction is direct and Ca(2+)-independent. Interacts with AHNAK2; the interaction is direct and Ca(2+)-independent.

Subcellular Location:

Cell membrane, sarcolemma; Single-pass type II membrane protein. Cytoplasmic vesicle membrane; Single-pass type II membrane protein (By similarity). Note=Colocalizes, during muscle differentiation, with BIN1 in the T-tubule system of myotubules and at the site of contact between two myotubes or a myoblast and a myotube. Wounding of myotubes led to its focal enrichment to the site of injury and to its relocalization in a Ca(2+)-dependent manner toward the plasma membrane. Colocalizes with AHNAK, AHNAK2 and PARVB at the sarcolemma of skeletal muscle. Detected on the apical plasma membrane of the syncytiotrophoblast. Reaches the plasmma membrane through a caveolin-independent mechanism. Retained by caveolin at the plasmma membrane (By similarity). Colocalizes, during muscle differentiation, with CACNA1S in the T-tubule system of myotubules (By similarity). Accumulates and colocalizes with fusion vesicles at the sarcolemma disruption sites (By similarity).

Tissue Specificity:

Expressed in skeletal muscle, myoblast, myotube and in the syncytiotrophoblast (STB) of the placenta (at protein level). Highly expressed in skeletal muscle. Also found in heart, brain, spleen, intestine, placenta and at lower levels in liver, lung, kidney and pancreas.

DISEASE:

Defects in DYSF are the cause of limb-girdle muscular dystrophy type 2B (LGMD2B) . LGMD2B is an autosomal recessive degenerative myopathy characterized by weakness and atrophy starting in the proximal pelvifemoral muscles, with onset in the late teens or later, massive elevation of serum creatine kinase levels and slow progression. Scapular muscle involvement is minor and not present at onset. Upper limb girdle involvement follows some years after the onset in lower limbs.

Similarity:

Belongs to the ferlin family.

Contains 5 C2 domains.

SWISS:

O75923

Gene ID:

8291

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

Dysferlin 是与浆膜内环境有关的一种跨膜蛋白，与肌膜损伤的修复有关.