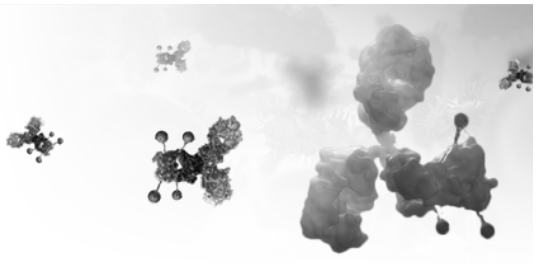

Human DYSF peptide (DAG-P0431)

This product is for research use only and is not intended for diagnostic use.

PRODUCT INFORMATION

Antigen Description	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]
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, kidne
Purity	70 - 90% by HPLC.
Conjugate	Unconjugated
Sequence Similarities	Belongs to the ferlin family.Contains 5 C2 domains.
Format	Liquid
Preservative	None
Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles. Information available upon request.

GENE INFORMATION

Gene Name	DYSF dysferlin [Homo sapiens (human)]
Official Symbol	DYSF

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Synonyms	DYSF; dysferlin; MMD1; FER1L1; LGMD2B; fer-1-like protein 1; dystrophy-associated fer-1-like 1; dystrophy-associated fer-1-like protein; limb girdle muscular dystrophy 2B (autosomal recessive); dysferlin, limb girdle muscular dystrophy 2B (autosomal recessive);
Entrez Gene ID	8291
mRNA Refseq	NM_001130455.1
Protein Refseq	NP_001123927.1
UniProt ID	O75923
Chromosome Location	2p13.3
Function	calcium ion binding; calcium-dependent phospholipid binding; phospholipid binding; protein binding;

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