

Recombinant Human DYSF Protein, MYC/DDK-tagged

Cat. No. DYSF-2773H Lot. No. (See product label)

SPECIFICATION

Product Overview	Recombinant Human DYSF protein, fused to MYC/DDK-tagged at C-terminus, was expressed in HEK293.
Species	Human
Source	HEK293
ProteinLength	1-2080 aa
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.
Form	25 mM Tris.HCl, pH 7.3, 100 mM glycine, 10% glycerol.
Molecular Mass	241.2 kDa
Purity	> 80% as determined by SDS-PAGE and Coomassie blue staining

 Tel: 1-631-559-9269 1-516-512-3133

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

Concentration >50 ug/mL as determined by microplate BCA method

GENE INFORMATION

Gene Name DYSF dysferlin, limb girdle muscular dystrophy 2B (autosomal recessive) [Homo sapiens]

Official Symbol DYSF

Synonyms FER1L1; LGMD2B; MMD1

Gene ID 8291

mRNA Refseq NM_001130455

Protein Refseq NP_001123927

MIM 603009

UniProt ID O75923

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