

Recombinant Human DYSF Protein, Myc/DDK-tagged, C13 and N15-labeled

Cat. No. DYSF-4000H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	DYSF MS Standard C13 and N15-labeled recombinant protein (NP_001124459) with a C-terminal MYC/DDK tag, was expressed in HEK293 cells.
Species	Human
Source	HEK293
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.
Molecular Mass	241.2 kDa
AA Sequence	MLCCLLVRASNLPSAKKDRRSDPVASLTFRGVKKRTKVIKNSVNPVWNEGFEDLK GIPLDQGSELHVVVKDHETMGRNRFLGEAKVPLREVLATPSLSASFNAPLLDTKKQP TGASLVLVQVSYTPLPGAVPLFPPTPLEPSPTLPDLVDVAGGGQSRAETWSLLSDST MDTRYSGKKWPAPTDGGEEDTEDQGLTGDEAEPFLDQSGGPGAPTTPRKLPSRP

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PPHYPGIKRKRSAPTSRKLLSDKPQDFQIRVQVIEGRQLPGVNIKPVVKVTAAGQTK
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LDKTESKIPARVVFQIWDNDKFSFDDFLGSLQLDLNRMPKPAKTAKKCSLDQLDDAF
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EPNMNPKLEDPRRPDTSFLWFTSPYKTMKFILWRRFRWAILFIILFILLFLAIFIYAFP
NYAAMKLVKPFSTRTRPLEQKLISEEDLAANDILDYKDDDDKV

Purity > 80% as determined by SDS-PAGE and Coomassie blue staining

Stability Stable for 3 months from receipt of products under proper storage and handling conditions.

Storage Store at -80 centigrade. Avoid repeated freeze-thaw cycles.

Concentration 50 µg/mL as determined by BCA

Storage Buffer 100 mM glycine, 25 mM Tris-HCl, pH 7.3.

GENE INFORMATION

Gene Name DYSF dysferlin [Homo sapiens (human)]

Official Symbol DYSF

Synonyms DYSF; dysferlin, limb girdle muscular dystrophy 2B (autosomal recessive); LGMD2B; dysferlin; FER1L1; fer-1-like protein 1; dystrophy-associated fer-1-like 1; dystrophy-associated fer-1-like protein; MMD1; FLJ00175; FLJ90168;

Gene ID 8291

mRNA Refseq NM_001130987

Protein Refseq NP_001124459

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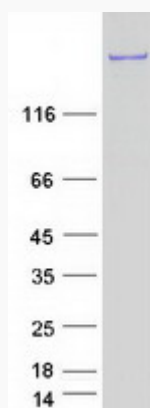
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MIM 603009

UniProt ID O75923

SDS-PAGE



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