

## Product datasheet for **SC326431**

### Dysferlin (DYSF) (NM\_001130978) Human Untagged Clone

#### Product data:

**Product Type:** Expression Plasmids  
**Product Name:** Dysferlin (DYSF) (NM\_001130978) Human Untagged Clone  
**Tag:** Tag Free  
**Symbol:** DYSF  
**Synonyms:** FER1L1; LGMD2B; LGMDR2; MMD1  
**Vector:** pCMV6 series  
**Fully Sequenced ORF:** >NCBI ORF sequence for NM\_001130978, the custom clone sequence may differ by one or more nucleotides

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 CTGGCCATCTTTCATCTACGCCTTCCCGAACTATGCTGCCATGAAGCTGGTGAAGCCCTTC  
 AGC

**Restriction Sites:**

Please inquire

**ACCN:**

NM\_001130978

**OTI Disclaimer:**

Our molecular clone sequence data has been matched to the reference identifier above as a point of reference. Note that the complete sequence of our molecular clones may differ from the sequence published for this corresponding reference, e.g., by representing an alternative RNA splicing form or single nucleotide polymorphism (SNP).

**OTI Annotation:**

This TrueClone is provided through our Custom Cloning Process that includes sub-cloning into OriGene's pCMV6 vector and full sequencing to provide a non-variant match to the expected reference without frameshifts, and is delivered as lyophilized plasmid DNA.

**Components:**

The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).

**Reconstitution Method:**

1. Centrifuge at 5,000xg for 5min.
2. Carefully open the tube and add 100ul of sterile water to dissolve the DNA.
3. Close the tube and incubate for 10 minutes at room temperature.
4. Briefly vortex the tube and then do a quick spin (less than 5000xg) to concentrate the liquid at the bottom.
5. Store the suspended plasmid at -20°C. The DNA is stable for at least one year from date of shipping when stored at -20°C.

**RefSeq:**
[NM\\_001130978.1](#), [NP\\_001124450.1](#)
**RefSeq Size:**

6953 bp

RefSeq ORF: 6306 bp

Locus ID: 8291

UniProt ID: [O75923](#)

Cytogenetics: 2p13.2

Protein Families: Transmembrane

**Gene Summary:** 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] Transcript Variant: This variant (11) differs in the 5' UTR and has multiple coding region differences, compared to variant 1. These differences cause translation initiation at an upstream AUG and a shorter isoform (11) with an alternate N-terminus compared to isoform 1.