

## Product datasheet

### Recombinant Human MBNL1 protein ab114825

[1 References](#) [1 Image](#)

#### Description

Product name	Recombinant Human MBNL1 protein
Expression system	Wheat germ
Accession	<b><u>AAH50535</u></b>
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	<p>           MGRCSRENCK YLHPPPHLKT QLEINGRNNL            IQQKNMAMLA QQMQLANAMM PGAPLQPVPM            FSVAPSLATN ASAAAFNPYL GPVSPSLVPA            EILPTAPMLV TGNPGVPVPA AAAAAAQKLM            RTDRLEVCRE YQRGNCNRGE NDCRFHPAD            STMIDTNDNT VTCMDYIKG RCSREKCKYF HPPAHLQAKI            KAAQYQVNQA AAAQAAATAA AMTQSAVKSL            KRPLEATFDL GIPQAVLPPL PKRPALEKTN GATAVNTGI            FQYQQALANM QLQQHTAFLP PGSILCMTPA            TSVVPMVHGA TPATVSAATT SATSVPFAAT ATANQIPIIS            AEHLTSHKYV TQM         </p>
Predicted molecular weight	64 kDa including tags
Amino acids	1 to 343
Tags	GST tag N-Terminus

#### Specifications

Our **Abpromise guarantee** covers the use of **ab114825** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	ELISA
	SDS-PAGE
	Western blot

Form	Liquid
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Additional notes	This product was previously labelled as Muscleblind-like 1.
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## Preparation and Storage

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### Stability and Storage

Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles.

pH: 8.00

Constituents: 0.3% Glutathione, 0.79% Tris HCl

## General Info

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### Function

Mediates pre-mRNA alternative splicing regulation. Acts either as activator or repressor of splicing on specific pre-mRNA targets. Inhibits cardiac troponin-T (TNNT2) pre-mRNA exon inclusion but induces insulin receptor (IR) pre-mRNA exon inclusion in muscle. Antagonizes the alternative splicing activity pattern of CELF proteins. Regulates the TNNT2 exon 5 skipping through competition with U2AF2. Inhibits the formation of the spliceosome A complex on intron 4 of TNNT2 pre-mRNA. Binds to the stem-loop structure within the polypyrimidine tract of TNNT2 intron 4 during spliceosome assembly. Binds to the 5'-YGCU(U/G)Y-3' consensus sequence. Binds to the IR RNA. Binds to expanded CUG repeat RNA, which folds into a hairpin structure containing GC base pairs and bulged, unpaired U residues.

### Tissue specificity

Highly expressed in cardiac, skeletal muscle and during myoblast differentiation. Weakly expressed in other tissues (at protein level). Expressed in heart, brain, placenta, lung, liver, skeletal muscle, kidney and pancreas.

### Involvement in disease

Plays a role in the pathogenesis of dystrophin myotonia type 1 (DM1) [MIM:160900]. A muscular disorder characterized by myotonia, muscle wasting in the distal extremities, cataract, hypogonadism, defective endocrine functions, male baldness and cardiac arrhythmias. Note=In muscle cells from DM1 patients, MBNL1 is sequestered by DMPK RNAs containing CUG triplet repeat expansions. MBNL1 binding is proportional to repeat length consistent with the direct correlation between the length of repeat expansion and disease severity.

### Sequence similarities

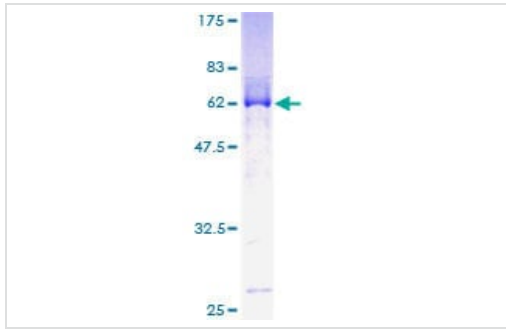
Belongs to the muscleblind family.  
Contains 4 C3H1-type zinc fingers.

### Cellular localization

Nucleus. Cytoplasm. Cytoplasmic granule. Localized with DDX1, TIAL1 and YBX1 in stress granules upon stress. Localized in the cytoplasm of multinucleated myotubes. Colocalizes with nuclear foci of retained expanded-repeat transcripts in myotubes from patients affected by myotonic dystrophy.

## Images

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12.5% SDS-PAGE Stained with Coomassie Blue

SDS-PAGE - Recombinant Human MBNL1 protein  
(ab114825)

**Please note:** All products are "FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC PROCEDURES"

### Our Abpromise to you: Quality guaranteed and expert technical support

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- Replacement or refund for products not performing as stated on the datasheet
- Valid for 12 months from date of delivery
- Response to your inquiry within 24 hours
- We provide support in Chinese, English, French, German, Japanese and Spanish
- Extensive multi-media technical resources to help you
- We investigate all quality concerns to ensure our products perform to the highest standards

If the product does not perform as described on this datasheet, we will offer a refund or replacement. For full details of the Abpromise, please visit <https://www.abcam.com/abpromise> or contact our technical team.

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