abcam

Product datasheet

# Recombinant Human MBNL1 protein ab114825

1 References　画像数 1

## 製品の詳細

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| 製品名 | Recombinant Human MBNL1 protein |
| 発現系 | Wheat germ |
| アクセッション番号 | AAH50535 |
| タンパク質長 | Full length protein |
| Animal free | No |
| 由来 | Recombinant |
| 生物種 | Human |
| 配列 | MGRCSRENCK YLHPPPHLKT QLEINGRNNL IQQKNMAMLA QQMQLANAMM PGAPLQPVPM FSVAPSLATN ASAAAFNPYL GPVSPSLVPA EILPTAPMLV TGNPGVPVPA AAAAAAQKLM RTDRLEVCRE YQRGNCNRGE NDCRFAHPAD STMIDTNDNT VTVCMDYIKG RCSREKCKYF HPPAHLQAKI KAAQYQVNQA AAAQAAATAA AMTQSAVKSL KRPLEATFDL GIPQAVLPPL PKRPALEKTN GATAVFNTGI FQYQQALANM QLQQHTAFLP PGSILCMTPA TSVVPMVHGA TPATVSAATT SATSVPFAAT ATANQIPIIS AEHLTSHKYV TQM |
| 予測される分子量 | 64 kDa including tags |
| 領域 | 1 to 343 |
| タグ | GST tag N-Terminus |

## 特性

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.

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| アプリケーション | ELISA |
| | SDS-PAGE |
| | Western blot |
| 製品の状態 | Liquid |

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| **備考** | This product was previously labelled as Muscleblind-like 1. |

## 前処理および保存

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| **保存方法および安定性** | Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles.<br>pH: 8.00<br>Constituents: 0.3% Glutathione, 0.79% Tris HCl |

## 関連情報

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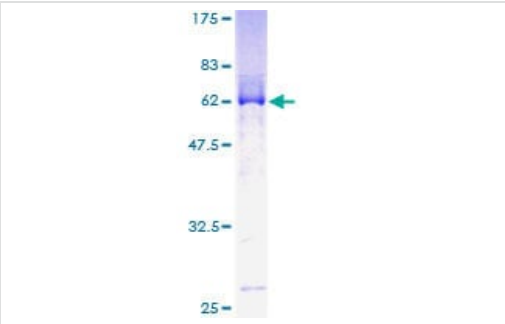
| **機能** | Mediates pre-mRNA alternative splicing regulation. Acts either as activator or repressor of splicing on specific pre-mRNA targets. Inhibits cardiac troponin-T (TNNT2) pre-mRNA exon inclusion but induces insulin receptor (IR) pre-mRNA exon inclusion in muscle. Antagonizes the alternative splicing 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. |
| **組織特異性** | 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. |
| **関連疾患** | Plays a role in the pathogenesis of dystrophia myotonica 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. |
| **配列類似性** | Belongs to the muscleblind family.<br>Contains 4 C3H1-type zinc fingers. |
| **細胞内局在** | 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. |

## 画像




SDS-PAGE - Recombinant Human MBNL1 protein (ab114825)

12.5% SDS-PAGE Stained with Coomassie Blue



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