

Product datasheet

Recombinant Human MBNL1 protein (Tagged)  
ab272043

1 Image

Description

<b>Product name</b>	Recombinant Human MBNL1 protein (Tagged)	
<b>Purity</b>	> 90 % SDS-PAGE.	
<b>Expression system</b>	Escherichia coli	
<b>Accession</b>	<a href="#">Q9NR56-5</a>	
<b>Protein length</b>	Full length protein	
<b>Animal free</b>	No	
<b>Nature</b>	Recombinant	
<b>Species</b>	Human	
<b>Sequence</b>	MAVSVTPIRDTKWLTLEVCREFQRGTC SRPDTECKFAHP SKSCQVENGRV IACFDSLKGRCSRENCKYLHPPPHLKTQLEINGRNNLIQQK NMAMLAQQM QLANAMMPGAPLQPVPMFSVAPSLATNASAAAFNPYLGP VSPSLVPAEIL PTAPMLVTGNPGVPVPA AAAAAAQAQLMRTDRLEVCREYQ RGNCNRGENDC RFAHPADSTMIDTNDNTVTVCM DYIKGRCSREKCKYFHPP AHLQAKIKAA QYQVNQAAAAQAAATAAAMGIPQAVLPPLPKRPALEKTN GATAVFNTGIF QYQQALANMQLQQHTAFLPPGSILCMT PATSVVPMVHGA TPATVSAATTS ATSVPF AATATANQIPIISAEHLTSHKYVTQM	
<b>Predicted molecular weight</b>	57 kDa including tags	
<b>Amino acids</b>	1 to 382	
<b>Additional sequence information</b>	N-terminal 6xHis-SUMO-tagged. Isoform 5.	

Specifications

Our [Abpromise guarantee](#) covers the use of **ab272043** in the following tested applications.

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

<b>Applications</b>	SDS-PAGE
<b>Form</b>	Liquid
<b>Additional notes</b>	Isoform 5.

## Preparation and Storage

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<b>Stability and Storage</b>	Shipped at 4°C. Upon delivery aliquot. Store at -20°C or -80°C. Avoid freeze / thaw cycle. Constituents: Tris buffer, PBS, Glycerol  5-50% glycerol
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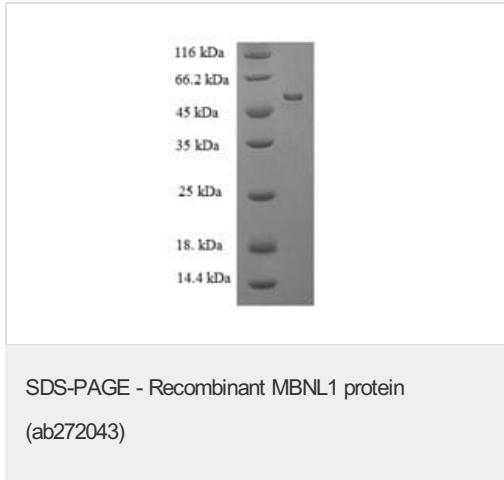
## General Info

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<b>Function</b>	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.
<b>Tissue specificity</b>	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.
<b>Involvement in disease</b>	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.
<b>Sequence similarities</b>	Belongs to the muscleblind family. Contains 4 C3H1-type zinc fingers.
<b>Cellular localization</b>	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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SDS-PAGE analysis of ab272043.

15% gel run under reducing conditions.

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

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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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