



Rabbit Anti-MBNL1 antibody

SL19111R

Product Name:	MBNL1
Chinese Name:	MBNL1蛋白抗体
Alias:	Muscleblind-like 1; EXP; EXP35; EXP40; EXP42; KIAA0428; MBNL; MBNL protein; MBNL1; MBNL1_HUMAN; Muscleblind 41kD isoform; Muscleblind like; Muscleblind like protein 1; Muscleblind like splicing regulator 1; Muscleblind-like protein 1; Triplet expansion RNA binding protein; Triplet-expansion RNA-binding protein.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Dog,Pig,Cow,Horse,Rabbit,Sheep,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	41kDa
Cellular localization:	The nucleuscytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human MBNL1:251-350/388
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	MBNL1 is a deduced 370 amino acid protein which is predominantly expressed in skeletal muscle, prostate, lung, heart, small intestine, ovary and placenta tissues. MBNL1 and MBNL2, which associate with ex-panded CUG repeats in vitro, both

localize to the nuclear foci in both DM1 and DM2 (myotonic dystrophy types 1 and 2), suggesting that the nuclear accumulation of mutant RNA is pathogenic in DM1, therefore implicating MBNL1 and 2 in the pathogenesis of both disorders.

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.

Subcellular Location:

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.

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.

DISEASE:

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.

Similarity:

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

SWISS:

Q9NR56

Gene ID:

4154

Database links:

[Entrez Gene: 4154](#) Human

[Entrez Gene: 425033](#) Chicken

[Entrez Gene: 781653](#) Cow

[Entrez Gene: 56758](#) Mouse

[Entrez Gene: 282635](#) Rat

[Omim: 606516](#) Human

[SwissProt: Q5ZKW9](#) Chicken

[SwissProt: Q9NR56](#) Human

[SwissProt: Q9JKP5](#) Mouse

[Unigene: 201858](#) Human

[Unigene: 725347](#) Human

[Unigene: 255723](#) Mouse

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.