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MBNL1 Rabbit mAb

Catalog # AP76580

Product Information

Application	WB
Primary Accession	Q9NR56
Reactivity	Human, Mouse
Host	Rabbit
Clonality	Monoclonal Antibody
Isotype	IgG
Conjugate	Unconjugated
Purification	Affinity Purified
Calculated MW	41817

Additional Information

Gene ID	4154
Other Names	MBNL1
Dilution	WB~~1:1000
Format	Liquid in 50mM Tris-Glycine(pH 7.4), 0.15M NaCl, 40%Glycerol, 0.01% sodium azide and 0.05% BSA.
Storage	Store at 4°C short term. Aliquot and store at -20°C long term. Avoid freeze/thaw cycles.

Protein Information

Name	MBNL1
Synonyms	EXP, KIAA0428, MBNL
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. Together with RNA binding proteins RBPMS and RBFOX2, activates vascular smooth muscle cells alternative splicing events (PubMed:37548402). Regulates NCOR2 alternative

	splicing (By similarity).
Cellular Location	Nucleus. Cytoplasm. Cytoplasmic granule. Note=Localized with DDX1, TIAL1 and YBX1 in stress granules upon stress (PubMed:18335541). Localized in the cytoplasm of multinucleated myotubes (PubMed:18335541). Colocalizes with nuclear foci of retained expanded-repeat transcripts in myotubes from patients affected by myotonic dystrophy (PubMed:10970838, PubMed:11590133, PubMed:11929853)
Tissue Location	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.

Background

This gene encodes a member of the muscleblind protein family which was initially described in Drosophila melanogaster. The encoded protein is a C3H-type zinc finger protein that modulates alternative splicing of pre-mRNAs. Muscleblind proteins bind specifically to expanded dsCUG RNA but not to normal size CUG repeats and may thereby play a role in the pathophysiology of myotonic dystrophy. Mice lacking this gene exhibited muscle abnormalities and cataracts. Several alternatively spliced transcript variants have been described but the full-length natures of only some have been determined. The different isoforms are thought to have different binding specificities and/or splicing activities.

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