MBNL1 蛋白抗体

产品货号： mlR19111

英文名称： MBNL1

中文名称： MBNL1 蛋白抗体

别　　名： 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.

研究领域： 细胞生物　转录调节因子

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Chicken, Dog, Pig, Cow, Horse, Rabbit, Sheep,

产品应用： ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500 （石蜡切片需做抗原修复）

not yet tested in other applications.

optimal dilutions/concentrations should be determined by the end user.

分 子 量： 41kDa

细胞定位： 细胞核 细胞浆

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human MBNL1:251-350/388

亚　　型： IgG

纯化方法： affinity purified by Protein A

储 存 液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： 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

产品介绍： 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 expanded 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

Important Note:

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