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RBM20 Rabbit pAb

Catalog # AP96267

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	Q5T481
Other Accession	Q5T481
Reactivity	Human, Mouse
Predicted	Rat, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	134258
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human RBM20
Epitope Specificity	361-460/1227
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Nucleus (By similarity).
SIMILARITY	Contains 1 RRM (RNA recognition motif) domain.
DISEASE	Defects in RBM20 are the cause of cardiomyopathy dilated type 1DD (CMD1DD) [MIM:613172]. A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a protein that likely binds RNA. Mutations in this gene have been associated with familial dilated cardiomyopathy. [provided by RefSeq, Mar 2010]

Additional Information

Gene ID	282996
Other Names	1110018J23Rik; 2010003H22Rik; RBM20_HUMAN; RBM20; RNA-binding motif protein 20; RBM20_MOUSE; RNA binding motif protein 20
Target/Specificity	Expressed in the heart.
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500,Flow-Cyt=1ug/test
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

Name	RBM20 {ECO:0000303|PubMed:22466703, ECO:0000312|HGNC:HGNC:27424}
Function	RNA-binding protein that acts as a regulator of mRNA splicing of a subset of genes encoding key structural proteins involved in cardiac development, such as TTN (Titin), CACNA1C, CAMK2D or PDLIM5/ENH (PubMed:22466703, PubMed:24960161, PubMed:26604136, PubMed:27496873, PubMed:27531932, PubMed:29895960, PubMed:30948719, PubMed:32840935, PubMed:34732726, PubMed:35427468). Acts as a repressor of mRNA splicing: specifically binds the 5'UCUU-3' motif that is predominantly found within intronic sequences of pre-mRNAs, leading to the exclusion of specific exons in target transcripts (PubMed:24960161, PubMed:30948719, PubMed:34732726). RBM20-mediated exon skipping is hormone-dependent and is essential for TTN isoform transition in both cardiac and skeletal muscles (PubMed:27531932, PubMed:30948719). RBM20-mediated exon skipping of TTN provides substrates for the formation of circular RNA (circRNAs) from the TTN transcripts (PubMed:27531932, PubMed:34732726). Together with RBM24, promotes the expression of short isoforms of PDLIM5/ENH in cardiomyocytes (By similarity).
Cellular Location	Nucleus {ECO:0000255|PROSITE-ProRule:PRU00130, ECO:0000269|PubMed:23886709, ECO:0000269|PubMed:30262925, ECO:0000269|PubMed:32840935, ECO:0000269|PubMed:33188278, ECO:0000269|PubMed:34732726, ECO:0000269|PubMed:35427468}. Cytoplasm, Cytoplasmic ribonucleoprotein granule. Note=The active form that regulates alternative splicing localizes to the nucleus (PubMed:33188278, PubMed:34732726, PubMed:35427468). Also localizes to cytoplasmic ribonucleoprotein granules; localization to cytoplasmic ribonucleoprotein granules plays an important regulatory role (PubMed:33188278, PubMed:34732726, PubMed:35427468). Subcellular localization is regulated by phosphorylation of different parts of the protein: while phosphorylation of the RS (arginine/serine-rich) region promotes nuclear localization, phosphorylation of the C-terminal disordered region promotes localization to cytoplasmic ribonucleoprotein granules (PubMed:35427468)
Tissue Location	Mainly expressed in the heart (PubMed:19712804, PubMed:23886709). Also expressed in skeletal muscle tissues, ovary, small intestine and colon (PubMed:23886709)

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