



Rabbit Anti-Human SMN2 Polyclonal Antibody (CABT-L2295)

This product is for research use only and is not intended for diagnostic use.

PRODUCT INFORMATION

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| Product Overview | Polyclonal Antibody to Survival Of Motor Neuron 2, Centromeric (Knockout Validated) |
| Specificity | The antibody is a rabbit polyclonal antibody raised against SMN2. It has been selected for its ability to recognize SMN2 in immunohistochemical staining and western blotting. |
| Target | SMN2 |
| Immunogen | Recombinant fragment corresponding to human SMN2 (Glu16~Pro251) |
| Isotype | IgG |
| Source/Host | Rabbit |
| Species Reactivity | Human, Mouse |
| Conjugate | Unconjugated |
| Applications | WB |
| Format | Liquid |
| Concentration | Lot specific |
| Size | 200 µg |
| Buffer | Supplied as solution form in 0.01M PBS with 50% glycerol, pH7.4. |
| Preservative | 0.05% Proclin-300 |
| Storage | Avoid repeated freeze/thaw cycles. Store at 4°C for frequent use. Aliquot and store at -20°C for 12 months. |

BACKGROUND

Introduction

This gene is part of a 500 kb inverted duplication on chromosome 5q13. This duplicated region contains at least four genes and repetitive elements which make it prone to rearrangements and deletions. The repetitiveness and complexity of the sequence have also caused difficulty in determining the organization of this genomic region. The telomeric and centromeric copies of this gene are nearly identical and encode the same protein. While mutations in the telomeric copy are associated with spinal muscular atrophy, mutations in this gene, the centromeric copy, do not lead to disease. This gene may be a modifier of disease caused by mutation in the telomeric copy. The critical sequence difference between the two genes is a single nucleotide in exon 7, which is thought to be an exon splice enhancer. Note that the nine exons of both the telomeric and centromeric copies are designated historically as exon 1, 2a, 2b, and 3-8. It is thought that gene conversion events may involve the two genes, leading to varying copy numbers of each gene. The full length protein encoded by this gene localizes to both the cytoplasm and the nucleus. Within the nucleus, the protein localizes to subnuclear bodies called gems which are found near coiled bodies containing high concentrations of small ribonucleoproteins (snRNPs). This protein forms heteromeric complexes with proteins such as SIP1 and GEMIN4, and also interacts with several proteins known to be involved in the biogenesis of snRNPs, such as hnRNP U protein and the small nucleolar RNA binding protein. Four transcript variants encoding distinct isoforms have been described. [provided by RefSeq, Sep 2008]

Keywords

SMNC;SMNT;SMA2;BCD541;SMA3;SMA4;SMA1;SMA

GENE INFORMATION

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| Gene Name | SMN2 survival of motor neuron 2, centromeric [Homo sapiens (human)] |
| Official Symbol | SMN2 |
| Synonyms | SMN2; survival of motor neuron 2, centromeric; SMNC; BCD541; GEMIN1; TDRD16B; C-BCD541; survival motor neuron protein; gemin-1; component of gems 1; tudor domain containing 16B; |
| Entrez Gene ID | 6607 |
| Protein Refseq | NP_059107 |
| UniProt ID | Q16637 |
| Chromosome Location | 5q13.2 |

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| Pathway | Gene Expression; Metabolism of non-coding RNA; RNA transport; Survival motor neuron (SMN) complex; snRNP Assembly; |
| Function | RNA binding; identical protein binding; protein binding; |