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SMN2

Reactivity: Human Mouse Rat

Tested applications:WB IHC

Recommended Dilution:WB 1:500 - 1:2000 IHC 1:50 - 1:200

Calculated MW:32kDa

Observed MW: Refer to figures

Immunogen:

Recombinant protein of human SMN2

Storage Buffer:

Store at -20. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 50% glycerol, pH7.3.

Synonym:

SMNC; BCD541; GEMIN1; TDRD16B; C-BCD541;

Catalog #:A1652

Antibody Type:

Polyclonal Antibody

Species: Rabbit

Gene ID:6607

Isotype: IgG

Swiss Prot:Q16637

Purity: Affinity purification

For research use only.

Background:

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.

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