

Recombinant Human SMN2 Protein, DYKDDDDK-tagged

Cat. No. SMN2-151H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human survival of motor neuron 2, centromeric(SMN2) protein, transcript variant b(NM_022876), with a DYKDDDDK tag, was expressed in human cells.
Species	Human
Source	Human Cells
Description	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

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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.

Form	Purified protein formulated in a sterile solution of TBS buffer, pH7.156, without any preservatives.
Molecular Mass	28.4 kDa
Endotoxin	Endotoxin level is < 0.1 ng/g of protein (<1EU /g)
Purity	>90% by SDS-PAGE gel and Coomassie Blue staining
Applications	Antigens, Western, ELISA and other in vitro binding or in vivo functional assays, and protein-protein interaction studies.

GENE INFORMATION

Gene Name	SMN2 survival of motor neuron 2, centromeric [Homo sapiens]
Official Symbol	SMN2
Synonyms	SMN2; survival of motor neuron 2, centromeric; survival motor neuron protein; BCD541; SMNC; gemin 1; gemin-1; component of gems 1; SMN1; C-BCD541; MGC5208; FLJ76644; MGC20996;
Gene ID	6607
mRNA Refseq	NM_017411

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Protein Refseq	NP_059107
MIM	601627
UniProt ID	Q16637

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