Recombinant Full Length Human SMN1 Protein

Cat. No. SMN1-506HF **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant full length Human Gemin 1, Isoform SMN-delta7 with N terminal proprietary tag, 56.76 kDa.
Species	Human
Source	In Vitro Cell Free System
ProteinLength	282 amino acids
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. However, mutations in this gene, the telomeric copy, are associated with spinal muscular atrophy; mutations in the centromeric copy do not lead to disease. The centromeric copy 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 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

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	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. Two transcript variants encoding distinct isoforms have been described.
Form	Liquid
Molecular Mass	56.760kDa inclusive of tags
AA Sequence	MAMSSGGSGGGVPEQEDSVLFRRGTGQSDDSDIWDDTALI KAYDKAVASFKHALK NGDICETSGKPKTTPKRKPAKKNKS QKKNTAASLQQWKVGDKCSAIWSEDGCIYPA TIASIDFKR ETCVVVYTGYGNREEQNLSDLLSPICEVANNIEQNAQENE NESQVSTD ESENSRSPGNKSDNIKPKSAPWNSFLPPPPPM PGPRLGPGKPGLKFNGPPPPPPP PPPHLLSCWLPPFPSGP PIIPPPPPICPDSLDDADALGSMLISWYMSGYHTGYYME M LA
Purity	Proprietary Purification
Storage	Shipped on dry ice. Upon delivery aliquot and store at -80 centigrade. Avoid freeze / thaw cycles.
Storage Buffer	pH: 8.00. Constituents:0.79% Tris HCl, 0.31% Glutathione.

GENE INFORMATION

Gene Name	SMN1 survival of motor neuron 1, telomeric [Homo sapiens]
Official Symbol	SMN1

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Synonyms	SMN1; survival of motor neuron 1, telomeric; SMA, SMA@, spinal muscular atrophy (Werdnig Hoffmann disease, Kugelberg Welander disease); survival motor neuron protein; BCD541; SMA1; SMA2; SMA3; SMNT
Gene ID	6606
mRNA Refseq	NM_000344
Protein Refseq	NP_000335
MIM	600354
UniProt ID	Q16637

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