

# Recombinant Full Length Human SMN1 Protein, C-Flag-tagged

Cat. No. SMN1-308HFL Lot. No. (See product label)

## SPECIFICATION

### Product Overview

Recombinant Full Length Human SMN1 Protein, fused to Flag-tag at C-terminus, was expressed in Mammalian cells.

### Species

Human

### Source

Mammalian 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. 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 found near coiled bodies containing high concentrations of small ribonucleoproteins (snRNPs). This protein forms heteromeric complexes with proteins such as SIP1 and

 Tel: 1-631-559-9269 1-516-512-3133

 Email: [info@creative-biomart.com](mailto:info@creative-biomart.com)  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

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. Multiple transcript variants encoding distinct isoforms have been described.

**Form** 25 mM Tris HCl, pH 7.3, 100 mM glycine, 10% glycerol.

**Molecular Mass** 31.7 kDa

**AA Sequence**  
MAMSSGGSGGGVPEQEDSVLFRRGTGQSDSDIWDDTALIKAYDKAVASFKHALK  
NGDICETSGKPKTTP KRKPAKKNKSQKKNTAASLQQWKVGDKCSAIWSEDGCIYPA  
TIASIDFKRETCVVVYTG YGNREEQNLS D LLSPICEVANNIEQNAQENENESQVSTD  
ESENSRSPGNKSDNIKPKSAPWNSFLPPPPMPGPRLGPGKP GLKFNGPPPPPPP  
PPPHLLSCWLPPFPSPGPIIPPPPICPDSLDDADALGSMLISWYMSGYHTGYMGMF  
RQNQKEGRCSHSLNTRTRPLEQKLISEEDLAANDILDYKDDDDKV

**Purity** > 80% as determined by SDS-PAGE and Coomassie blue staining.

**Stability** Stable for 12 months from the date of receipt of the product under proper storage and handling conditions. Avoid repeated freeze-thaw cycles.

**Storage** Store at -80 centigrade.

**Concentration** >50 ug/mL as determined by microplate BCA method.

**Preparation** Recombinant protein was captured through anti-DDK affinity column followed by conventional chromatography steps.

**Protein Families** Druggable Genome, Stem cell - Pluripotency

**Full Length** Full L.

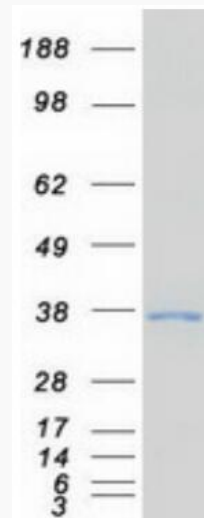
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## GENE INFORMATION

<b>Gene Name</b>	SMN1 survival of motor neuron 1, telomeric [ Homo sapiens (human) ]
<b>Official Symbol</b>	SMN1
<b>Synonyms</b>	SMA; SMN; SMA1; SMA2; SMA3; SMA4; SMA@; SMNT; BCD541; GEMIN1; TDRD16A; T-BCD541
<b>Gene ID</b>	6606
<b>mRNA Refseq</b>	NM_000344.4
<b>Protein Refseq</b>	NP_000335.1
<b>MIM</b>	600354
<b>UniProt ID</b>	Q16637



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Coomassie blue staining of purified SMN1 protein.

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