Recombinant Human SMN1 Protein, His-tagged

Cat. No. SMN1-458H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human SMN1(Ala2~Asn294) fused with His tag at N-terminal was expressed in E. coli.
Species	Human
Source	E.coli
ProteinLength	Ala2~Asn294
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. Multiple transcript variants encoding distinct isoforms have been described.
Form	PBS, pH7.4, containing 0.01% SKL, 1mM DTT, 5% Trehalose and Proclin300.
Molecular Mass	35.4kDa
Endotoxin	<1.0EU per 1ug (determined by the LAL method)
Purity	> 95%
Applications	Positive Control; Immunogen; SDS-PAGE; WB. If bio-activity of the protein is needed, please check active protein
Stability	The thermal stability is described by the loss rate. The loss rate was determined by accelerated thermal degradation test, that is, incubate the protein at 37 centigrade for 48h, and no obvious degradation and precipitation were observed. The loss rate is less than 5% within the expiration date under appropriate storage condition.
Storage	Avoid repeated freeze/thaw cycles. Store at 2-8 centigrade for one month. Aliquot and store at -80 centigrade for 12 months.
Reconstitution	Reconstitute in PBS or others

GENE INFORMATION

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

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Official Symbol	SMN1
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; gemin 1; gemin-1; component of gems 1; SMA; SMN; SMA4; SMA@; SMN2; T-BCD541;
Gene ID	6606
mRNA Refseq	NM_000344
Protein Refseq	NP_000335
MIM	600354
UniProt ID	Q16637

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