

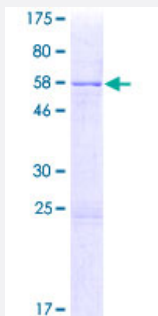
Full-Length

## SMN2 (Human) Recombinant Protein (P01)

Catalog # H00006607-P01

Size 25 ug, 10 ug

### Applications



### Specification

#### Product Description

Human SMN2 full-length ORF ( NP\_075013.1, 1 a.a. - 282 a.a.) recombinant protein with GST-tag at N-terminal.

#### Sequence

MAMSSGGSGGGVPEQEDSVLFRRGTGQSDDSDWDDTALIKAYDKAVASFHALKNGDICETSG  
KPKTTPKRKPAKKNKSQKKNTAASLQQWKVGDKCSAWSEDGCMPTIASIDFKRETCVVVYTG  
YGNREEQNLSDLLSPICEVANNIEQNAQENENESQVSTDESENSRSPGNKSDNIKPKSAPWNSFL  
PPPPMPGPRLGPGKPLKFNGPPPPPPPPHLLSCWLPPFPSPGPIIPPPPPICPDSLDDAD  
ALGSMLISWYMSGYHTGYMEMLA

#### Host

Wheat Germ (in vitro)

#### Theoretical MW (kDa)

56.9

#### Preparation Method

[in vitro wheat germ expression system](#)

#### Purification

Glutathione Sepharose 4 Fast Flow

#### Quality Control Testing

12.5% SDS-PAGE Stained with Coomassie Blue.

#### Storage Buffer

50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

#### Storage Instruction

Store at -80°C. Aliquot to avoid repeated freezing and thawing.

#### Note

Best use within three months from the date of receipt of this protein.

## Applications

- Enzyme-linked Immunoabsorbent Assay
- Western Blot (Recombinant protein)
- Antibody Production
- Protein Array

## Gene Info — SMN2

Entrez GeneID [6607](#)

GeneBank Accession# [NM\\_022875.1](#)

Protein Accession# [NP\\_075013.1](#)

Gene Name SMN2

Gene Alias BCD541, C-BCD541, FLJ76644, MGC20996, MGC5208, SMNC

Gene Description survival of motor neuron 2, centromeric

Omim ID [601627](#)

Gene Ontology [Hyperlink](#)

### Gene Summary

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. [provided by RefSeq]

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**Other Designations**OTTHUMP00000125236|OTTHUMP00000125237|gemin 1

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## Disease

- [Amyotrophic lateral sclerosis](#)
- [Disease Progression](#)
- [Genetic Predisposition to Disease](#)
- [Muscular Atrophy](#)
- [Nerve Degeneration](#)
- [Spinal Muscular Atrophies of Childhood](#)
- [Spinal muscular atrophy](#)