

SMN1 (human), (recombinant) (His- tag)

Survival Motor Neuron (SMN) is a ~38 kDa protein produced chiefly by the SMN1 gene, located on the telomeric portion of chromosome 5q. A nearly identical centromeric copy of the gene (SMN2) also produces a small amount of full-length SMN protein, but due to a translationally silent C(R)T transition that results in alternative splicing of the pre-mRNA, most of the resulting SMN is truncated, causing reduced protein stability and lower overall SMN levels. Deletion or mutation of the SMN1 gene results in a reduced level of full-length SMN protein and manifests as a range of neuromuscular phenotypes in humans as the disease spinal muscular atrophy (SMA). SMA is characterized by muscle weakness and atrophy, functional disability and is the most common lethal genetic disease of infants and toddlers. Approximately one in 35 adults is a carrier of the SMN1 mutation. The incidence of SMA is 1 in 6,000 to 1 in 10,000 live births. SMN protein is present in the cell cytoplasm, and also in the nucleus where it is concentrated in "gem" structures associated with Cajal bodies. SMN protein is a constituent of Gemin-containing complexes, and is thought to participate in many aspects of RNA metabolism. SMN complexes have been shown to mediate the assembly of uridine-rich small nuclear ribonucleoproteins (snRNPs), which in turn act as critical components of spliceosomes.

Citations: 8

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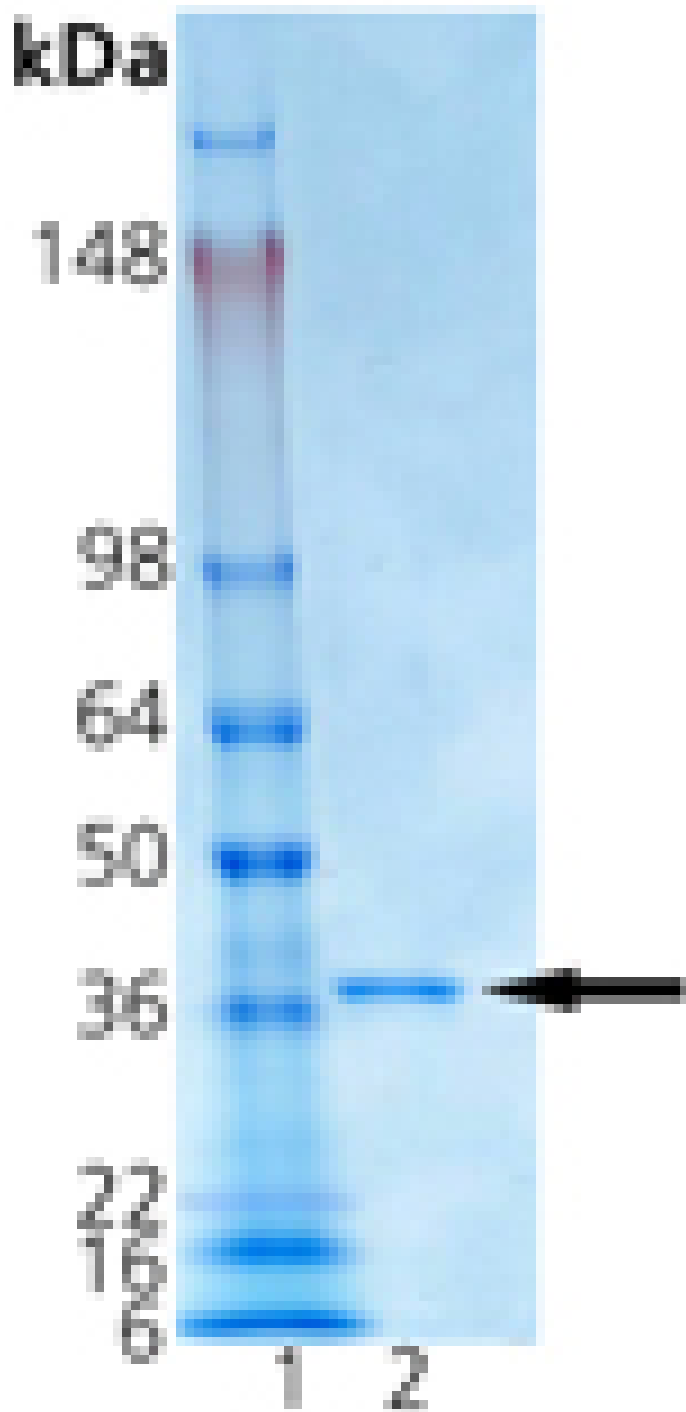
Ordering Information

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ADI-NBP-201-050	50µg
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Manuals, SDS & CofA

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Handling & Storage

Long Term Storage -80°C

Shipping Dry Ice

Regulatory Status RUO - Research Use Only

Product Details

Alternative Name Survival Motor Neuron

Formulation Liquid. In 20 mM Tris HCl pH 7.4, 300 mM potassium chloride and 10% glycerol.

MW 40kDa

Purity ≥90% (SDS-PAGE)

Purity Detail Purified by multi-step chromatography.

Source Produced in *E. coli*. The N-terminus is fused with a His tag.

Technical Info / Product Notes Protected by US Patent no. US. 6,080,577.

UniProt ID Q16637



ENZO LIFE SCIENCES,
INC.
Phone: 800.942.0430
[info-
usa@enzolifesciences.com](mailto:info-usa@enzolifesciences.com)

European Sales Office
ENZO LIFE SCIENCES
(ELS) AG
Phone: +41 61 926 8989
[info-
eu@enzolifesciences.com](mailto:info-eu@enzolifesciences.com)

Belgium, The Netherlands
& Luxembourg
Phone: +32 3 466 0420
[info-
be@enzolifesciences.com](mailto:info-be@enzolifesciences.com)

France
Phone: +33 472 440 655
[info-
fr@enzolifesciences.com](mailto:info-fr@enzolifesciences.com)

Germany
Phone: +49 7621 5500 526
[info-
de@enzolifesciences.com](mailto:info-de@enzolifesciences.com)

UK & Ireland
Phone (UK customers):
0845 601 1488
Phone: +44 1392 825900
[info-
uk@enzolifesciences.com](mailto:info-uk@enzolifesciences.com)