

Product datasheet

Recombinant Human SMN/Gemin 1 protein ab114802

[1 Image](#)

Overview

Product name	Recombinant Human SMN/Gemin 1 protein
Protein length	Full length protein

Description

Nature	Recombinant
Source	Wheat germ

Amino Acid Sequence

Accession	Q16637-3
Species	Human
Sequence	MAMSSGGSGGGVPEQEDSVLFRRGTGQSDSDIWDDTALIKAYDKAVASF KHALKNGDICETSGKPKTTPKRKPAKKNKSQKKNTAASLQQWKVGDKCSA WSEDGCYPATIASIDFKRETCVVVYTYGYNREEQNLSDLLSPICEVAN NIEQNAQENENESQVSTDESENSRSPGNKSDNIKPKSAPWNSFLPPPPPM PGPRLGPGKPLKFNGLPPPPPPPPHLLSCWLPPFPSPGPIIPPPPIC PDSLDDADALGSMLISWYMSGYHTGYMEMLA

Molecular weight	57 kDa including tags
Amino acids	1 to 282

Specifications

Our [Abpromise guarantee](#) covers the use of **ab114802** in the following tested applications.

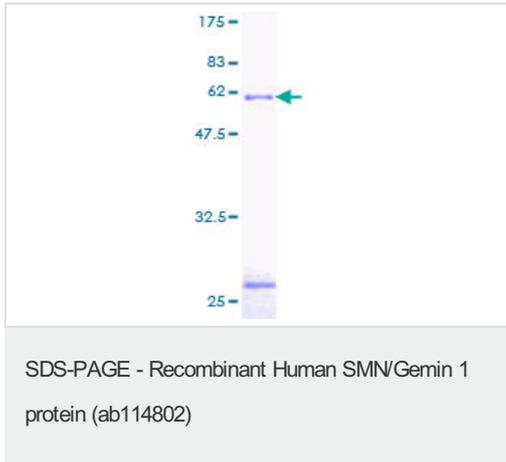
The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	Western blot SDS-PAGE ELISA
Form	Liquid
Additional notes	Protein concentration is above or equal to 0.05 µg/ul. Best used within three months from the date of receipt.

Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.3% Glutathione, 0.79% Tris HCl
General Info	
Function	The SMN complex plays an essential role in spliceosomal snRNP assembly in the cytoplasm and is required for pre-mRNA splicing in the nucleus. It may also play a role in the metabolism of snoRNPs.
Tissue specificity	Expressed in a wide variety of tissues. Expressed at high levels in brain, kidney and liver, moderate levels in skeletal and cardiac muscle, and low levels in fibroblasts and lymphocytes. Also seen at high levels in spinal cord. Present in osteoclasts and mononuclear cells (at protein level).
Involvement in disease	<p>Defects in SMN1 are the cause of spinal muscular atrophy autosomal recessive type 1 (SMA1) [MIM:253300]. Spinal muscular atrophy refers to a group of neuromuscular disorders characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7-skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit.</p> <p>Defects in SMN1 are the cause of spinal muscular atrophy autosomal recessive type 2 (SMA2) [MIM:253550]. SMA2 is an autosomal recessive spinal muscular atrophy of intermediate severity, with onset between 6 and 18 months. Patients do not reach the motor milestone of standing, and survive into adulthood.</p> <p>Defects in SMN1 are the cause of spinal muscular atrophy autosomal recessive type 3 (SMA3) [MIM:253400]. SMA3 is an autosomal recessive spinal muscular atrophy with onset after 18 months. SMA3 patients develop ability to stand and walk and survive into adulthood.</p> <p>Defects in SMN1 are the cause of spinal muscular atrophy autosomal recessive type 4 (SMA4) [MIM:271150]. SMA4 is an autosomal recessive spinal muscular atrophy characterized by symmetric proximal muscle weakness with onset in adulthood and slow disease progression. SMA4 patients can stand and walk.</p>
Sequence similarities	Belongs to the SMN family. Contains 1 Tudor domain.
Cellular localization	Cytoplasm. Nucleus > gem. Localized in subnuclear structures next to coiled bodies, called Gemini of Cajal bodies.

Images



ab114802 on 12.5% SDS-PAGE Stained with Coomassie Blue.

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