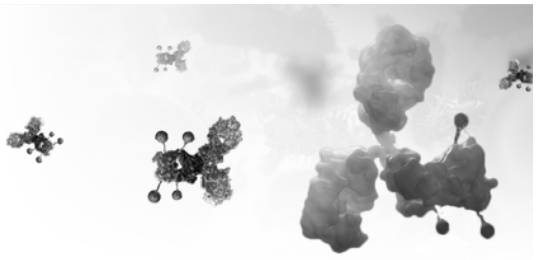

Rabbit Anti-Human SMN1 Polyclonal Antibody (CABT-L2287)

This product is for research use only and is not intended for diagnostic use.

PRODUCT INFORMATION

Product Overview	Polyclonal Antibody to Survival Of Motor Neuron 1, Telomeric (Knockout Validated)
Specificity	The antibody is a rabbit polyclonal antibody raised against SMN1. It has been selected for its ability to recognize SMN1 in immunohistochemical staining and western blotting.
Target	SMN1
Immunogen	Recombinant fragment corresponding to human SMN1 (Ala2~Asn294)
Isotype	IgG
Source/Host	Rabbit
Species Reactivity	Human, Pig
Purification	Antigen-specific affinity chromatography followed by Protein A affinity chromatography
Conjugate	Unconjugated
Applications	WB
Format	Liquid
Concentration	Lot specific
Size	200 µg
Buffer	Supplied as solution form in 0.01M PBS with 50% glycerol, pH7.4.
Preservative	0.05% Proclin-300

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Storage	Avoid repeated freeze/thaw cycles. Store at 4°C for frequent use. Aliquot and store at -20°C for 12 months.
Ship	4°C with ice bags

BACKGROUND

Introduction	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 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. [provided by RefSeq, Jul 2014]
Keywords	BCD541;SMA1;SMA2;SMA3;SMA4;SMNT;T-BCD541;SMNC;Gemin-1;Component of gems 1;Spinal Muscular Atrophy(Werdnig-Hoffmann Disease,Kugelberg-Welander Disease)

GENE INFORMATION

Gene Name	SMN1 survival of motor neuron 1, telomeric [Homo sapiens (human)]
Official Symbol	SMN1
Synonyms	SMN1; survival of motor neuron 1, telomeric; SMA; SMN; SMA1; SMA2; SMA3; SMA4; SMA@; SMNT; BCD541; GEMIN1; TDRD16A; T-BCD541; survival motor neuron protein; gemin-1; component of gems 1; tudor domain containing 16A; survival motor neuron 1 protein;
Entrez Gene ID	6606
Protein Refseq	NP_000335

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UniProt ID	Q16637
Chromosome Location	5q13.2
Pathway	Gene Expression; Metabolism of non-coding RNA; RNA transport; Survival motor neuron (SMN) complex; snRNP Assembly;
Function	RNA binding; identical protein binding; protein binding;

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