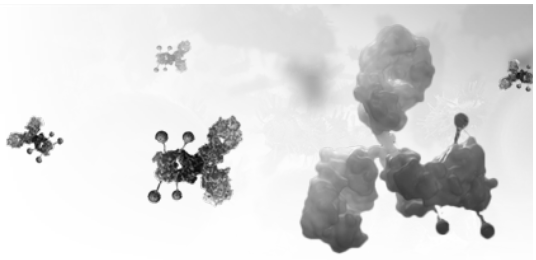

Human SMN1 blocking peptide (CDBP2737)

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

PRODUCT INFORMATION

Product Overview	Blocking/Immunizing peptide for anti-SMN1/SMN2 antibody
Antigen 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 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. Two transcript variants encoding distinct isoforms have been described. [provided by RefSeq, Sep 2008]
Species	Human
Conjugate	Unconjugated
Applications	Apuri, BL, ELISA
Format	Lyophilized powder
Size	100 µg

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Preservative	None
Storage	Shipped at ambient temperature, store at -20°C.

GENE INFORMATION

Gene Name	SMN1 survival of motor neuron 1, telomeric [Homo sapiens]
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;
Entrez Gene ID	6606
mRNA Refseq	NM_000344
Protein Refseq	NP_000335
UniProt ID	Q16637
Chromosome Location	5q13.2
Pathway	Gene Expression, organism-specific biosystem; Metabolism of non-coding RNA, organism-specific biosystem; RNA transport, organism-specific biosystem; RNA transport, conserved biosystem; Survival motor neuron (SMN) complex, organism-specific biosystem; snRNP Assembly, organism-specific biosystem;

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