

## Product datasheet for **TA303208**

### SMN1 Goat Polyclonal Antibody

#### Product data:

Product Type:	Primary Antibodies
Applications:	WB
Recommended Dilution:	ELISA: 1:1,000. WB: 2-4µg/ml.
Reactivity:	Human
Host:	Goat
Isotype:	IgG
Clonality:	Polyclonal
Immunogen:	Peptide with sequence C-DESENSRSPGNKSDN, from the internal region of the protein sequence according to NP_075012.1; NP_000335.1; NP_075013.1; NP_075014.1; NP_075015.1; NP_059107.1.
Formulation:	Supplied at 0.5 mg/ml in Tris saline, 0.02% sodium azide, pH7.3 with 0.5% bovine serum albumin.
Concentration:	lot specific
Purification:	Purified from goat serum by ammonium sulphate precipitation followed by antigen affinity chromatography using the immunizing peptide. Supplied at 0.5 mg/ml in Tris saline, 0.02% sodium azide, pH7.3 with 0.5% bovine serum albumin. Aliquot and store at -20°C. Minimize freezing and thawing.
Conjugation:	Unconjugated
Storage:	Store at -20°C as received.
Stability:	Stable for 12 months from date of receipt.
Gene Name:	survival of motor neuron 1, telomeric
Database Link:	<a href="#">NP_000335</a> <a href="#">Entrez Gene 6606 Human</a> <a href="#">Q16637</a>



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**Background:**

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 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 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 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]

**Synonyms:**

BCD541; GEMIN1; SMA; SMA1; SMA2; SMA3; SMA4; SMA@; SMN; SMNT; T-BCD541; TDRD16A

**Protein Families:**

Druggable Genome, Stem cell - Pluripotency

**Product images:**

TA303208 (2ug/ml) staining of Human Brain (Cerebellum) lysate (35ug protein in RIPA buffer). Primary incubation was 1 hour. Detected by chemiluminescence.