10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

Goat Anti-SMN1 / SMN2 Antibody
Peptide-affinity purified goat antibody
Catalog # AF2008a

Specification

Goat Anti-SMN1 / SMN2 Antibody - Product Information

Application	**WB, E**
Primary Accession	Q16637
Other Accession	NP_059107, 6606, 6607
Reactivity	**Human**
Host	**Goat**
Clonality	**Polyclonal**
Concentration	**0.5 mg/ml**
Isotype	**IgG**
Calculated MW	**31849**

Goat Anti-SMN1 / SMN2 Antibody - Additional Information

Gene ID 6606;6607

Other Names
Survival motor neuron protein, Component of gems 1, Gemin-1, SMN1, SMN, SMNT

Dilution
WB~~1:1000
E~~N/A

Format
0.5 mg IgG/ml in Tris saline (20mM Tris pH7.3, 150mM NaCl), 0.02% sodium azide, with 0.5% bovine serum albumin

Storage
Maintain refrigerated at 2-8°C for up to 6 months. For long term storage store at -20°C in small aliquots to prevent freeze-thaw cycles.

Precautions
Goat Anti-SMN1 / SMN2 Antibody is for research use only and not for use in diagnostic or therapeutic procedures.

Goat Anti-SMN1 / SMN2 Antibody - Protein Information

Name SMN1

Synonyms SMN, SMNT

Function
The SMN complex catalyzes the assembly of small nuclear ribonucleoproteins (snRNPs), the

building blocks of the spliceosome, and thereby plays an important role in the splicing of cellular pre- mRNAs (PubMed:18984161, PubMed:9845364). Most spliceosomal snRNPs contain a common set of Sm proteins SNRPB, SNRPD1, SNRPD2, SNRPD3, SNRPE, SNRPF and SNRPG that assemble in a heptameric protein ring on the Sm site of the small nuclear RNA to form the core snRNP (Sm core) (PubMed:18984161). In the cytosol, the Sm proteins SNRPD1, SNRPD2, SNRPE, SNRPF and SNRPG are trapped in an inactive 6S pICln-Sm complex by the chaperone CLNS1A that controls the assembly of the core snRNP (PubMed:18984161). To assemble core snRNPs, the SMN complex accepts the trapped 5Sm proteins from CLNS1A forming an intermediate (PubMed:18984161). Within the SMN complex, SMN1 acts as a structural backbone and together with GEMIN2 it gathers the Sm complex subunits (PubMed:17178713, PubMed:21816274, PubMed:22101937). Binding of snRNA inside 5Sm ultimately triggers eviction of the SMN complex, thereby allowing binding of SNRPD3 and SNRPB to complete assembly of the core snRNP (PubMed:31799625). Ensures the correct splicing of U12 intron- containing genes that may be important for normal motor and proprioceptive neurons development (PubMed:23063131). Also required for resolving RNA-DNA hybrids created by RNA polymerase II, that form R- loop in transcription terminal regions, an important step in proper transcription termination (PubMed:26700805). May also play a role in the metabolism of small nucleolar ribonucleoprotein (snoRNPs).

Cellular Location
Nucleus, gem. Nucleus, Cajal body. Cytoplasm. Cytoplasmic granule. Perikaryon. Cell projection, neuron projection. Cell projection, axon {ECO:0000250|UniProtKB:P97801}. Cytoplasm, myofibril, sarcomere, Z line {ECO:0000250|UniProtKB:P97801}. Note=Colocalizes with actin and at the Z-line of skeletal muscle (By similarity). Under stress conditions colocalizes with RPP20/POP7 in punctuated cytoplasmic granules (PubMed:14715275). Colocalized and redistributed with ZPR1 from the cytoplasm to nuclear gems (Gemini of coiled bodies) and Cajal bodies (PubMed:11283611). Colocalizes with FMR1 in cytoplasmic granules in the soma and neurite cell processes (PubMed:18093976) {ECO:0000250|UniProtKB:P97801, ECO:0000269|PubMed:11283611, ECO:0000269|PubMed:14715275, ECO:0000269|PubMed:18093976}

Tissue Location
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).

Goat Anti-SMN1 / SMN2 Antibody - Protocols

Provided below are standard protocols that you may find useful for product applications.

- Western Blot
- Blocking Peptides
- Dot Blot
- Immunohistochemistry
- Immunofluorescence
- Immunoprecipitation

- Flow Cytomety
- Cell Culture

Goat Anti-SMN1 / SMN2 Antibody - Images

AF2008a (1 µg/ml) staining of Human Cerebellum lysate (35 µg protein in RIPA buffer). Primary incubation was 1 hour. Detected by chemiluminescence.

Goat Anti-SMN1 / SMN2 Antibody - 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 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.

Goat Anti-SMN1 / SMN2 Antibody - References

[Study of SMN gene in Chinese children with spinal muscular atrophy] Liu WL, et al. Zhongguo Dang Dai Er Ke Za Zhi, 2010 Jul. PMID 20637152. SMN, Gemin2 and Gemin3 associate with beta-actin mRNA in the cytoplasm of neuronal cells in vitro. Todd AG, et al. J Mol Biol, 2010 Sep 3. PMID 20620147. Splicing regulation of the survival motor neuron genes and implications for treatment of spinal muscular atrophy. Bebee TW, et al. Front Biosci, 2010 Jun 1. PMID 20515750. Molecular characterization of SMN copy number derived from carrier screening and from core families with SMA in a Chinese population. Sheng-Yuan Z, et al. Eur J Hum Genet, 2010 Sep. PMID 20442745. Large-scale population carrier screening for spinal muscular atrophy in Israel--effect of ethnicity on the false-negative rate. Sukenik-Halevy R, et al. Genet Test Mol Biomarkers, 2010 Jun. PMID 20373854.