Immunotag™ SMN1 Monoclonal Antibody

Antibody Specification

Catalog No.	ITM0588
Product Description	Immunotag™ SMN1 Monoclonal Antibody
Size	50 µg, 100 µg
Conjugation	HRP, Biotin, FITC, Alexa Fluor® 350, Alexa Fluor® 405, Alexa Fluor® 488, Alexa Fluor® 555, Alexa Fluor® 594, Alexa Fluor® 647
IMPORTANT NOTE	This product is custom manufactured with a lead time of 3-4 weeks. Once in production, this item cannot be cancelled from an order and is not eligible for return.
Target Protein	SMN1
Clonality	Monoclonal
Storage/Stability	-20°C/1 year
Application	WB,IHC-p,IF,ELISA
Recommended Dilution	Western Blot: 1/500 - 1/2000. Immunohistochemistry: 1/200 - 1/1000. Immunofluorescence: 1/200 - 1/1000. ELISA: 1/10000. Not yet tested in other applications.
Concentration	1 mg/ml
Reactive Species	Human,Monkey
Host Species	Mouse
Immunogen	Purified recombinant fragment of human SMN1 expressed in E. Coli.
Specificity	SMN1 Monoclonal Antibody detects endogenous levels of SMN1 protein.
Purification	Affinity purification
Form	Ascitic fluid containing 0.03% sodium azide.
Gene Name	SMN1
Accession No.	Q16637 P97801
Alternate Names	SMN1; SMN; SMNT; SMN2; SMNC; Survival motor neuron protein; Component of gems 1; Gemin-1

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Description	survival of motor neuron 1, telomeric(SMN1) Homo sapiens 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 des
Protein Expression	Amygdala,Epithelium,Fetal brain,Kidney,Lung,Placenta,Prostate,Skele
Subcellular Localization	nucleus,nucleoplasm,cytoplasm,cytosol,Cajal body,Z disc,SMN complex,SMN-Sm protein complex,cytoplasmic ribonucleoprotein granule,neuron projection,perikaryon,Gemini of coiled bodies,

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Protein Function	Experimental confirmation may be lacking for some isoforms,disease: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.,disease: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.,disease: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.,disease: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.,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.,miscellaneous:The SMN gene is present in two highly homologous and functional copies (TelSMN/SMN1 and CenSMN/SMN2). The telomeric copy of SMN gene (TelSMN/SMN1) seems to be the SMA-determining gene while the centromeric copy seems unaffected.,online information:The Singapore human mutation and polymorphism database,similarity:Belongs to the SMN family.,similarity:Contains 1 Tudor domain.,subcellular location:Localized in subnuclear structures next to coiled bodies, called Gemini of Cajal bodies (Gems).,subunit:Component of an import snRNP complex composed of KPNB1, RNUT1, SMN1 and ZNF259. Part of the core SMN complex that contains SMN1, SIP1/GEMIN2, DDX20/GEMIN3, GEMIN4, GEMIN5, GEMIN6, GEMIN7, GEMIN8 and STRAP/UNRIP. Interacts with DDX20, FBL, NOLA1, RNUT1, SYNCRIP and with several spliceosomal snRNP core Sm proteins, including SNRPB, SNRPD1, SNRPD2, SNRPD3, SNRPE and ILF3. Interacts with OSTF1.,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).,
Usage	For Research Use Only! Not for diagnostic or therapeutic procedures.