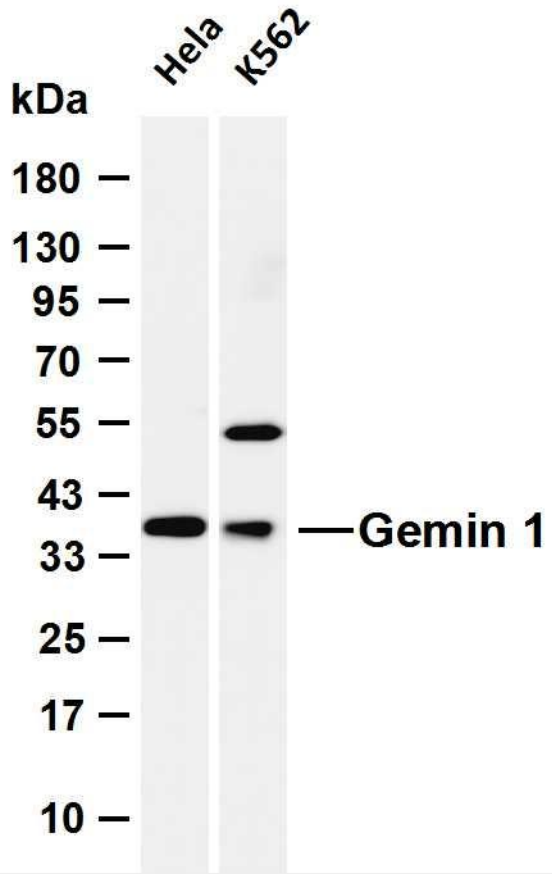
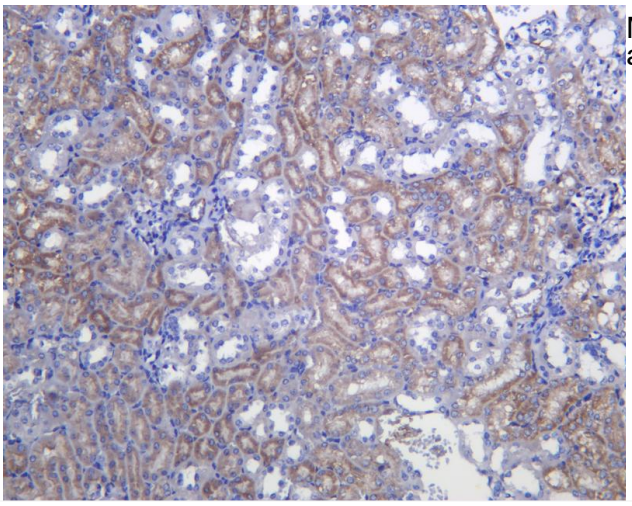
Gemin 1 Rabbit mAb

Catalog No	YP-rAb-16954
Isotype	IgG
Reactivity	Human,Mouse,Rat
Applications	WB,IHC,IF,IP,ELISA
Gene Name	SMN1;SMN2
Protein Name	Survival motor neuron protein
Purification Process	Protein A
Specificity	Endogenous
Formulation	PBS, 50% glycerol, 0.05% Proclin 300, 0.05%BSA
Source	Monoclonal, Rabbit,IgG
Dilution	IHC 1:200-1:500; WB 1:1000-1:20000; IF 1:200-1:1000; ELISA 1:5000-1:20000; IP 1:50-1:200; Note: For IHC, we suggest antigen retrieval with TE buffer pH 9.0
Concentration	0.5 mg/ml
Purity	≥90%
Storage Stability	-15° C to -25° C/1 year(Do not lower than -25° C)
Synonyms	SMN1 ; SMN ; SMNT ; SMN2 ; SMNC ; Survival motor neuron protein ; Component of gems 1 ; Gemin-1
Observed Band	38kD
Calculated Molecular Weight	32kD
Cell Pathway	Nucleus, gem . Nucleus, Cajal body . Cytoplasm . Cytoplasmic granule . Perikaryon . Cell projection, neuron projection . Cell projection, axon . Cytoplasm, myofibril, sarcomere, Z line . 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). .
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).
Function	Alternative products: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).,
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 des
matters needing attention	Avoid repeated freezing and thawing!
Usage suggestions	This product can be used in immunological reaction related experiments. For more information, please consult technical personnel.



Various whole cell lysates were separated by 4-20% SDS-PAGE, and the membrane was blotted with anti-Gemin 1 antibody. The HRP-conjugated Goat anti-Rabbit IgG (H + L) antibody was used to detect the antibody. Lane 1: Hela Lane 2: K562 Predicted band size: 32kDa Observed band size: 38kDa



Mouse kidney was stained with anti-Gemin 1 Rabbit antibody