

SMN1 polyclonal antibody

Catalog # PAB12734 Size 100 ug

Applications



Western Blot (Cell lysate)

The cell lysate derived from HeLa was immunoprobed by SMN1 polyclonal antibody (Cat # PAB12734) at 1 : 500. An immunoreactive band is observed around ~35 kDa (1). This band is abolished by pre-incubation with immunizing peptide (2).

Specification	
Product Description	Rabbit polyclonal antibody raised against synthetic peptide of SMN1.
Immunogen	A synthetic peptide corresponding to C-terminus of human SMN1.
Host	Rabbit
Theoretical MW (kDa)	35
Reactivity	Human
Specificity	This antibody recognizes ~35 KDa of human SMN1.
Form	Liquid
Purification	Affinity purification
Recommend Usage	ELISA (0.01-0.1 ug/mL) Immunohistochemistry (2-5 ug/mL) Immunoprecipitation (2-5 ug/mL) Western Blot (0.1-1 ug/mL) The optimal working dilution should be determined by the end user.
Storage Buffer	In PBS, pH 7.2 (Antibody Stabilizer).

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Product Information

Storage Instruction

Store at 2-8°C for three months without detectable loss of activity. For long term storage store at -20° C to -80°C. Aliquot to avoid repeated freezing and thawing.

Applications

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- Immunohistochemistry
- Immunoprecipitation
- Enzyme-linked Immunoabsorbent Assay

Gene Info — SMN1

Entrez GenelD	<u>6606</u>
Gene Name	SMN1
Gene Alias	BCD541, SMA, SMA1, SMA2, SMA3, SMA4, SMA@, SMN, SMNT, T-BCD541
Gene Description	survival of motor neuron 1, telomeric
Omim ID	<u>253300 253400 253550 271150 600354</u>
Gene Ontology	Hyperlink



Gene Summary

Product Information

This gene is part of a 500 kb inverted duplication on chromosome 5q13. This duplicated region c ontains at least four genes and repetitive elements which make it prone to rearrangements and d eletions. The repetitiveness and complexity of the sequence have also caused difficulty in determi ning the organization of this genomic region. The telomeric and centromeric copies of this gene a re nearly identical and encode the same protein. However, mutations in this gene, the telomeric c opy, are associated with spinal muscular atrophy; mutations in the centromeric copy do not lead t o disease. The centromeric copy may be a modifier of disease caused by mutation in the telomer ic 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 c onversion 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, t he protein localizes to subnuclear bodies called gems which are found near coiled bodies contain ing high concentrations of small ribonucleoproteins (snRNPs). This protein forms heteromeric co mplexes with proteins such as SIP1 and GEMIN4, and also interacts with several proteins known t o 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. [provide d by RefSeq

Other Designations

OTTHUMP00000125198|gemin 1

Publication Reference

• Gemin2 plays an important role in stabilizing the survival of motor neuron complex.

Ogawa C, Usui K, Aoki M, Ito F, Itoh M, Kai C, Kanamori-Katayama M, Hayashizaki Y, Suzuki H. The Journal of Biological Chemistry 2007 Feb; 282(15):11122.

Disease

- Acute Disease
- <u>Amyotrophic lateral sclerosis</u>
- <u>Chronic Disease</u>
- Disease Progression
- Genetic Predisposition to Disease
- <u>Muscular Atrophy</u>
- Nerve Degeneration
- Spinal Muscular Atrophies of Childhood
- Spinal muscular atrophy