

Product datasheet for TA343297

Gemin 1 (SMN2) Rabbit Polyclonal Antibody

Product data:

Product Type: Primary Antibodies

Applications: WB

Recommended Dilution: WB

Reactivity: Human

Host: Rabbit

Isotype: IgG

Clonality: Polyclonal

Immunogen: The immunogen for anti-SMN2 antibody is: synthetic peptide directed towards the N-terminal

region of SMN2. Synthetic peptide located within the following region: KHALKNGDICETSGKPKTTPKRKPAKKNKSQKKNTAASLQQWKVGDKCSA

Formulation: Liquid. Purified antibody supplied in 1x PBS buffer with 0.09% (w/v) sodium azide and 2%

sucrose.

Conjugation: Unconjugated

Storage: Store at -20°C as received.

Stability: Stable for 12 months from date of receipt.

Predicted Protein Size: 27 kDa

Gene Name: survival of motor neuron 2, centromeric

Database Link: NP 075015

Entrez Gene 6607 Human

Q16637



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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 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. While mutations in the telomeric copy are associated with spinal muscular atrophy, mutations in this gene, the centromeric copy, do not lead to disease. This gene 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 full length 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. Four transcript variants encoding distinct isoforms have been described.

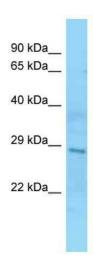
Synonyms: BCD541; C-BCD541; GEMIN1; SMNC; TDRD16B

Note: Immunogen Sequence Homology: Human: 100%; Dog: 93%; Horse: 92%; Pig: 86%; Rabbit:

85%; Goat: 82%; Mouse: 79%

Protein Families: Druggable Genome

Product images:



WB Suggested Anti-SMN2 Antibody; Titration: 1.0 ug/ml; Positive Control: Jurkat Whole Cell SMN2 is supported by BioGPS gene expression data to be expressed in Jurkat