

CAB9707

Product Information

Product SKU:	CAB9707	Gene ID:	6606	Size:	20uL, 100uL
Clone No:	ARC1710	Host Species:	Rabbit	Reactivity:	Human,Rat

Additional Information

Observed MW:	38kDa	Conjugate:	Unconjugated
Calculated MW:	32kDa	Isotype:	IgG

Immunogen Information

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. Multiple transcript variants encoding distinct isoforms have been described.

Recommended Dilution: WB,1:500 - 1:1000 IHC-P,1:50 - 1:200

Synonyms: SMA; SMN; SMA1; SMA2; SMA3; SMA4; SMA@; SMNT; BCD541; GEMIN1; TDRD16A; T-BCD541; SMN1

Purification Method: Affinity purification

Immunogen: Recombinant fusion protein containing a sequence corresponding to amino acids 103-200 of human SMN1 (Q16637).

Contact Details | Dublin, Ireland

Email: techsupport@assaygenie.com | **Web:** www.assaygenie.com

Copyright © 2020 Reagent Genie, All Rights Reserved. All information / detail is correct at time of going to print.

Storage:

Store at -20°C. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 0.05% BSA, 50% glycerol, pH 7.3.

Contact Details | Dublin, Ireland

Email: techsupport@assaygenie.com | **Web:** www.assaygenie.com

Copyright © 2020 Reagent Genie, All Rights Reserved. All information / detail is correct at time of going to print.