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Product datasheet for SC335049

SMN1 (NM_001297715) Human Untagged Clone

Product data:

Product Type: Expression Plasmids

Product Name: SMN1 (NM_001297715) Human Untagged Clone

Tag: Tag Free

Symbol: SMN1

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

Vector: pCMV6 series

Fully Sequenced ORF: >NCBI ORF sequence for NM_001297715, the custom clone sequence may differ by one or more nucleotides

ATGGCGATGAGCAGCGGCGGCAGTGGTGGCGGCGTCCCGGAGCAGGAGGATTCCGTGCTGTTCCGGCGCG
GCACAGGCCAGAGCGATGATTCTGACATTTGGGATGATACAGCACTGATAAAAGCATATGATAAAGCTGT
GGCTTCATTTAAGCATGCTCTAAAGAATGGTGACATTTGTGAAACTTCGGGTAAACCAAAAACCACACCT
AAAAGAAAACCTGCTAAGAAGAATAAAAGCCAAAAGAAGAATACTGCAGCTTCCTTACAACAGTGGAAAG
TTGGGGACAAATGTTCTGCCATTTGGTCAGAAGACGGTTGCATTTACCCAGCTACCATTGCTTCAATTGA
TTTTAAGAGAGAAACCTGTGTTGTGGTTTACACTGGATATGGAAATAGAGAGGAGCAAAATCTGTCCGAT
CTACTTTCCCCAATCTGTGAAGTAGCTAATAATATAGAACAAAATGCTCAAGAGAATGAAAATGAAAGCC
AAGTTTCAACAGATGAAAGTGAGAACTCCAGGTCTCCTGGAAATAAATCAGATAACATCAAGCCCAAATC
TGCTCCATGGAACTCTTTTCTCCCTCCACCACCCCCCATGCCAGGGCCAAGACTGGGACCAGGAAAGCCA
GGTCTAAAATTCAATGGCCCACCACCGCCACCGCCACCACCACCACCCCACTTACTATCATGCTGGCTGC
CTCCATTTCCTTCTGGACCACCAATAATTCCCCCACCACCTCCCATATGTCCAGATTCTCTTGATGATGC
TGATGCTTTGGGAAGTATGTTAATTTCATGGTACATGAGTGGCTATCATACTGGCTATTATATGGAAATG
CTGGCATAG

Restriction Sites: SgfI-MluI

ACCN: NM_001297715

OTI Disclaimer: Our molecular clone sequence data has been matched to the reference identifier above as a point of reference. Note that the complete sequence of our molecular clones may differ from the sequence published for this corresponding reference, e.g., by representing an alternative RNA splicing form or single nucleotide polymorphism (SNP).

Components: The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).

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Reconstitution Method:
1. Centrifuge at 5,000xg for 5min.
2. Carefully open the tube and add 100ul of sterile water to dissolve the DNA.
3. Close the tube and incubate for 10 minutes at room temperature.
4. Briefly vortex the tube and then do a quick spin (less than 5000xg) to concentrate the liquid at the bottom.
5. Store the suspended plasmid at -20°C. The DNA is stable for at least one year from date of shipping when stored at -20°C.

RefSeq: NM_001297715.1, NP_001284644.1

RefSeq Size: 1571 bp

RefSeq ORF: 849 bp

Locus ID: 6606

UniProt ID: Q16637

Cytogenetics: 5q13.2

Protein Families: Druggable Genome, Stem cell - Pluripotency

Gene Summary: 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. [provided by RefSeq, Jul 2014]
Transcript Variant: This variant (a) lacks the penultimate exon, which results in an alternate translation stop codon, compared to variant d. The resulting isoform (a) is shorter and has a distinct C-terminus, compared to isoform d. Sequence Note: The RefSeq transcript and protein were derived from genomic sequence to make the sequence consistent with the reference genome assembly. The genomic coordinates used for the transcript record were based on alignments.