

Product datasheet for SC112583

Gemin 1 (SMN2) (NM 017411) Human Untagged Clone

Product data:

Product Type: Expression Plasmids

Product Name: Gemin 1 (SMN2) (NM_017411) Human Untagged Clone

Tag: Tag Free
Symbol: Gemin 1

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

Mammalian Cell

Selection:

None

Vector: pCMV6-XL5

E. coli Selection: Ampicillin (100 ug/mL)

Fully Sequenced ORF: >OriGene sequence for NM_017411 edited

GGCCCACGCTGCGCACCCGCGGGTTTGCTATGGCGATGAGCAGCGGCGGCAGTGGTGGC GGCGTCCCGGAGCAGGAGGATTCCGTGCTGTTCCGGCGCGCACAGGCCAGAGCGATGAT TCTGACATTTGGGATGATACAGCACTGATAAAAGCATATGATAAAGCTGTGGCTTCATTT AAGCATGCTCTAAAGAATGGTGACATTTGTGAAACTTCGGGTAAACCAAAAACCACACCT AAAAGAAAACCTGCTAAGAAGAATAAAAGCCAAAAGAAGAATACTGCAGCTTCCTTACAA CAGTGGAAAGTTGGGGACAAATGTTCTGCCATTTGGTCAGAAGACGGTTGCATTTACCCA GCTACCATTGCTTCAATTGATTTTAAGAGAGAAACCTGTGTTGTGGTTTACACTGGATAT GGAAATAGAGAGGAGCAAAATCTGTCCGATCTACTTTCCCCAATCTGTGAAGTAGCTAAT AATATAGAACAGAATGCTCAAGAGAATGAAAATGAAAGCCAAGTTTCAACAGATGAAAGT GAGAACTCCAGGTCTCCTGGAAATAAATCAGATAACATCAAGCCCAAATCTGCTCCATGG AACTCTTTTCTCCCTCCACCACCCCCCATGCCAGGGCCAAGACTGGGACCAGGAAAGCCA TGCTGGCTGCCTCCATTTCCTTCTGGACCACCAATAATTCCCCCACCACCACCTCCCATATGT CCAGATTCTCTTGATGATGCTGATGCTTTGGGAAGTATGTTAATTTCATGGTACATGAGT CATTCCTTAAATTAAGGAGAAATGCTGGCATAGAGCAGCACTAAATGACACCACTAAAGA AACGATCAGACAGATCTGGAATGTGAAGCGTTATAGAAGATAACTGGCCTCATTTCTTCA AAATATCAAGTGTTGGGAAAGAAAAAGGAAGTGGAATGGGTAACTCTTCTTGATTAAAA GTTATGTAATAACCAAATGCAATGTGAAATATTTTACTGGACTCTATTTTGAAAAACCAT CTGTAAAAGACTGAGGTGGGGGTGGGAGGCCAGCACGGTGGTGAGGCAGTTGAGAAAATT GAAGGGTGTTGTAGTTTATAAAAGACTGTCTTAATTTGCATACTTAAGCATTTAGGAATG TGGCAAAATGTTACAGAATCTAACTGGTGGACATGGCTGTTCATTGTACTGTTTTTTTCT AAAAAAAAAAAAAAAAAAAAA

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Restriction Sites: ECORI-NOT

ACCN: NM_017411

Insert Size: 1500 bp

OTI Disclaimer: Due to the inherent nature of this plasmid, standard methods to replicate additional amounts

of DNA in E. coli are highly likely to result in mutations and/or rearrangements. Therefore, OriGene does not guarantee the capability to replicate this plasmid DNA. Additional amounts of DNA can be purchased from OriGene with batch-specific, full-sequence verification at a reduced cost. Please contact our customer care team at customercom or by

calling 301.340.3188 option 3 for pricing and delivery.

The molecular sequence of this clone aligns with the gene accession number as a point of reference only. However, individual transcript sequences of the same gene can differ through naturally occurring variations (e.g. polymorphisms), each with its own valid existence. This clone is substantially in agreement with the reference, but a complete review of all prevailing variants is recommended prior to use. More info

OTI Annotation: The ORF of this clone has been fully sequenced and found to be a perfect match to

NM_017411.2.

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).

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: <u>NM 017411.2</u>, <u>NP 059107.1</u>

 RefSeq Size:
 1623 bp

 RefSeq ORF:
 885 bp

 Locus ID:
 6607

 UniProt ID:
 Q16637

 Cytogenetics:
 5q13.2

Protein Families: Druggable Genome



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. 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. [provided by RefSeq, Sep 2008] Transcript Variant: This variant (d) represents the longest transcript and encodes the longest isoform (d).