

Product datasheet for SC207586

Gemin 1 (SMN2) (NM 022877) Human 3' UTR Clone

Product data:

Product Type: 3' UTR Clones

Product Name: Gemin 1 (SMN2) (NM_022877) Human 3' UTR Clone

Symbol: Gemin 1

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

Mammalian Cell

Selection:

Neomycin

Vector: pMirTarget (PS100062)

ACCN: NM_022877

Insert Size: 598 bp

Insert Sequence: >SC207586 3'UTR clone of NM_022877

The sequence shown below is from the reference sequence of NM_022877. The complete

sequence of this clone may contain minor differences, such as SNPs.

Blue=Stop Codon Red=Cloning site

GGCAAGTTGGACGCCCGCAAGATCCGCGAGATTCTCATTAAGGCCAAGAAGGGCGGAAAGATCGCCGTG

TAACAATTGGCAGAGCTCAGAATTCAAGCGATCGCC

CGAGATTTCGATTCCACCGCCGCCTTCTATGAAAGG

Restriction Sites: Sgfl-Mlul

OTI Disclaimer: Our molecular clone sequence data has been matched to the sequence identifier above as a

point of reference. Note that the complete sequence of this clone is largely the same as the

reference sequence but may contain minor differences, e.g., single nucleotide

polymorphisms (SNPs).



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Components:

The cDNA clone is shipped in a 2-D bar-coded Matrix tube as 10 ug dried plasmid DNA. The package also includes 100 pmols of both the corresponding 5' and 3' vector primers in separate vials.

RefSeq:

NM 022877.2

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 RefSeg, Sep 2008]

Locus ID: 6607 **MW:** 23.4