

Product datasheet for RC204374

Gemin 1 (SMN2) (NM_017411) Human Tagged ORF Clone

Product data:

Product Type: Expression Plasmids Product Name: Gemin 1 (SMN2) (NM 017411) Human Tagged ORF Clone Tag: Myc-DDK Gemin 1 Symbol: Synonyms: BCD541; C-BCD541; GEMIN1; SMNC; TDRD16B Mammalian Cell Neomycin Selection: Vector: pCMV6-Entry (PS100001) E. coli Selection: Kanamycin (25 ug/mL) **ORF** Nucleotide >RC204374 representing NM_017411 Red=Cloning site Blue=ORF Green=Tags(s) Sequence: TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC GCCGCGATCGCC

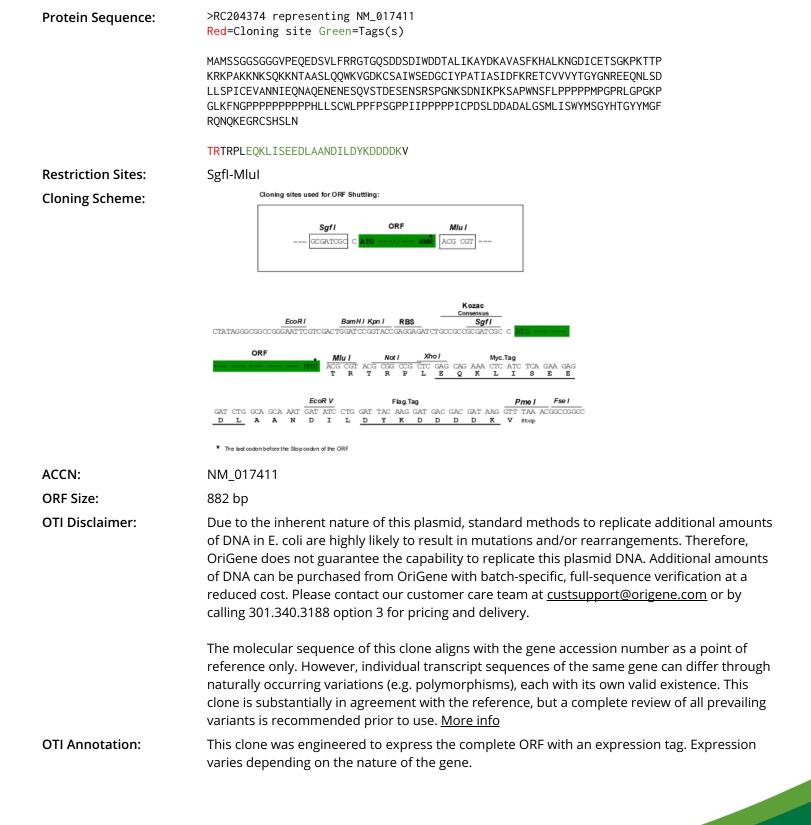
ACGCGTACGCGGCCGCTCGAGCAGAAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCCTGGATT ACAAGGATGACGACGATAAG**GTTTAA**



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| Gemin 1 (SMN2) (NM_017411) Human Tagged ORF Clone – RC204374 | |
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| 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: | Centrifuge at 5,000xg for 5min. Carefully open the tube and add 100ul of sterile water to dissolve the DNA. Close the tube and incubate for 10 minutes at room temperature. Briefly vortex the tube and then do a quick spin (less than 5000xg) to concentrate the liquid at the bottom. 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. |
| Note: | Plasmids are not sterile. For experiments where strict sterility is required, filtration with 0.22um filter is required. |
| RefSeq: | <u>NM 017411.4</u> |
| RefSeq Size: | 1634 bp |
| RefSeq ORF: | 885 bp |
| Locus ID: | 6607 |
| UniProt ID: | <u>Q16637</u> |
| Cytogenetics: | 5q13.2 |
| Protein Families: | Druggable Genome |
| MW: | 31.8 kDa |
| 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 |

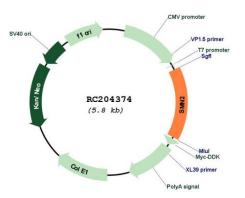
and the small nucleolar RNA binding protein. Four transcript variants encoding distinct isoforms have been described. [provided by RefSeq, Sep 2008]

several proteins known to be involved in the biogenesis of snRNPs, such as hnRNP U protein

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Product images:



Circular map for RC204374

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