

Product datasheet for RC201623

Gemin 1 (SMN2) (NM_022875) Human Tagged ORF Clone

Product data:

Product Type: Expression Plasmids Product Name: Gemin 1 (SMN2) (NM_022875) 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 >RC201623 ORF sequence Red=Cloning site Blue=ORF Green=Tags(s) Sequence: TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC GCCGCGATCGCC

ACGCGTACGCGGCCGCTCGAGCAGAAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCCTGGATT ACAAGGATGACGACGATAAGGTTTAA

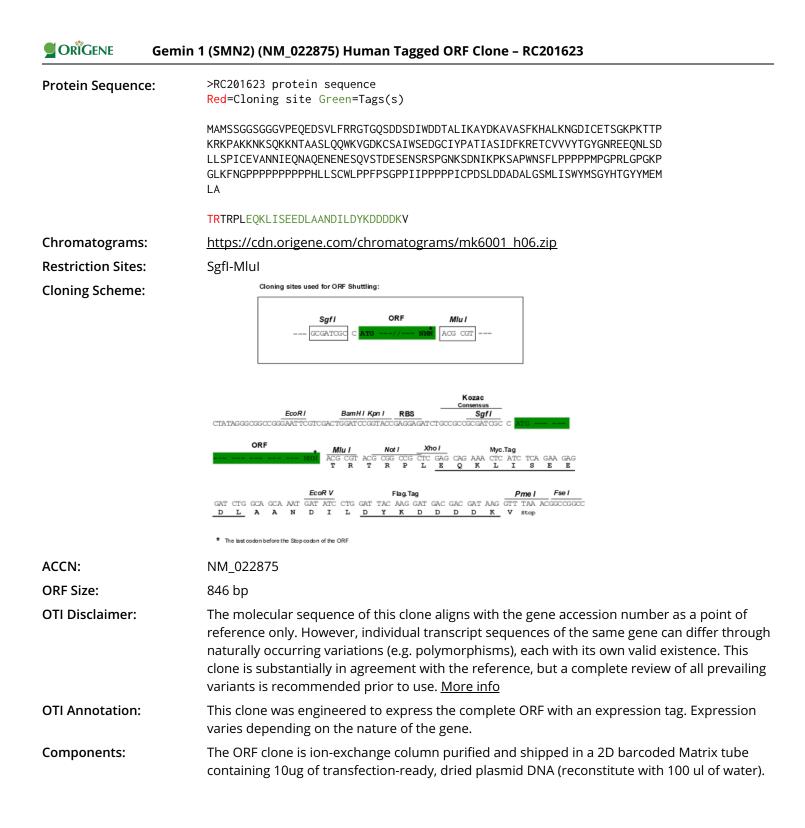


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Reconstitution Method:	 Centrifuge at 5,000xg for 5min. Carefully open the tube and add 100ul of sterile water to dissolve the DNA. Close the tube and incubate for 10 minutes at room temperature. Briefly vortex the tube and then do a quick spin (less 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.
RefSeq:	<u>NM 022875.1, NP 075013.1</u>
RefSeq Size:	1580 bp
RefSeq ORF:	849 bp
Locus ID:	6607
UniProt ID:	<u>Q16637</u>
Cytogenetics:	5q13.2
Protein Families:	Druggable Genome
MW:	30.5 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

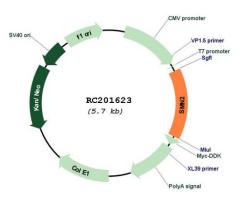
the two genes is a single nucleotide in exon 7, which is thought to be an exon splice enhancer. Note that the nine exons of both the telomeric and centromeric copies are designated historically as exon 1, 2a, 2b, and 3-8. It is thought that gene conversion events may involve the two genes, leading to 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 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]

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Product images:



Circular map for RC201623

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