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Product datasheet for SC308807

Gemin 1 (SMN2) (NM_022877) Human Untagged Clone

Product data:

Product Type:	Expression Plasmids
Product Name:	Gemin 1 (SMN2) (NM_022877) 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_022877 edited GGCCCCACGCTGCGCACCCGCGGGTTTGCTATGGCGATGAGCAGCGGCGGCAGTGGTGGC GGCGTCCCGGAGCAGGAGGATTCCGTGCTGTTCCGGCGCGGCACAGGCCAGAGCGATGAT TCTGACATTTGGGATGATACAGCACTGATAAAAGCATATGATAAAGCTGTGGCTTCATTT AAGCATGCTCTAAAGAATGGTGACATTTGTGAAACTTCGGGTAAACCAAAAACCACACCT AAAAGAAAACCTGCTAAGAAGAATAAAAGCCAAAAGAAGAATACTGCAGCTTCCTTACAA CAGTGGAAAGTTGGGGACAAATGTTCTGCCATTTGGTCAGAAGACGGTTGCATTTACCCA GCTACCATTGCTTCAATTGATTTTAAGAGAGAAACCTGTGTTGTGGTTTACACTGGATAT GGAAATAGAGAGGAGCAAAATCTGTCCGATCTACTTTCCCCAATCTGTGAAGTAGCTAAT AATATAGAACAGAATGCTCAAGAGAATGAAAATGAAAGCCAAGTTTCAACAGATGAAAGT GAGAACTCCAGGTCTCCTGGAAATAAATCAGATAACATCAAGCCCAAATCTGCTCCATGG AACTCTTTTCTCCCTCCACCACCCCCCATGCCAGGGCCAAGACTGGGACCAGGAAAGATA ATTCCCCCACCACCTCCCATATGTCCAGATTCTCTTGATGATGCTGATGCTTTGGGAAGT ATGTTAATTTCATGGTACATGAGTGGCTATCATACTGGCTATTATATGGAAATGCTGGCA TAG
Restriction Sites:	Please inquire
ACCN:	NM_022877
Insert Size:	800 bp
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).

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OTI Annotation:	The ORF of this clone has been fully sequenced and found to be a perfect match to NM_022877.1.
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:	NM_022877.1, NP_075015.1
RefSeq Size:	1473 bp
RefSeq ORF:	753 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 (c) lacks two alternate exons in the 3' CDS compared to variant d. The resulting protein (isoform c) is shorter and has a distinct C-terminus compared to isoform d.