

## Product datasheet for TP321156M

### Gemin 1 (SMN2) (NM\_022876) Human Recombinant Protein

#### Product data:

Product Type:	Recombinant Proteins
Description:	Recombinant protein of human survival of motor neuron 2, centromeric (SMN2), transcript variant b, 100 µg
Species:	Human
Expression Host:	HEK293T
Expression cDNA Clone or AA Sequence:	>RC221156 representing NM_022876 <b>Red</b> =Cloning site <b>Green</b> =Tags(s)
	MAMSSGGSGGGVPEQEDSVLFRRGTGQSDSDIWDDETALIKAYDKAVASFKHALKNGDICETSGKPKTTP KRKPAKKNKSQKNTAASLQQWKVGDKCSAIWSEDGCIYPATIASIDFKRETCVVVYTYGYNREEQNLSL LLSPICEVANNIEQNAQENENESQVSTDESENSRSPGNKSDNIKPKSAPWNSFLPPPPMPGPRLGPGKI IPPPPICPDSLDDADALGSMLISWYMSGYHTGYMGRQNQKEGRCSHSLN
	<b>TRTRPLEQKLISEEDLAANDILDYKDDDDKV</b>
Tag:	C-Myc/DDK
Predicted MW:	28.4 kDa
Concentration:	>0.05 µg/µL as determined by microplate BCA method
Purity:	> 80% as determined by SDS-PAGE and Coomassie blue staining
Buffer:	25 mM Tris-HCl, 100 mM glycine, pH 7.3, 10% glycerol
Preparation:	Recombinant protein was captured through anti-DDK affinity column followed by conventional chromatography steps.
Note:	For testing in cell culture applications, please filter before use. Note that you may experience some loss of protein during the filtration process.
Storage:	Store at -80°C.
Stability:	Stable for 12 months from the date of receipt of the product under proper storage and handling conditions. Avoid repeated freeze-thaw cycles.
RefSeq:	<a href="#">NP_075014</a>
Locus ID:	6607



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UniProt ID: [Q16637](#)

RefSeq Size: 1527

Cytogenetics: 5q13.2

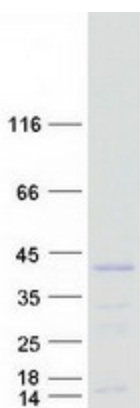
RefSeq ORF: 786

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

**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]

**Protein Families:** Druggable Genome

### Product images:



Coomassie blue staining of purified SMN2 protein (Cat# [TP321156]). The protein was produced from HEK293T cells transfected with SMN2 cDNA clone (Cat# [RC221156]) using MegaTran 2.0 (Cat# [TT210002]).