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Product datasheet for PH321156

Gemin 1 (SMN2) (NM_022876) Human Mass Spec Standard

Product data:

Product Type:	Mass Spec Standards
Description:	SMN2 MS Standard C13 and N15-labeled recombinant protein (NP_075014)
Species:	Human
Expression Host:	HEK293
Expression cDNA Clone or AA Sequence:	RC221156
Predicted MW:	28.4 kDa
Protein Sequence:	>RC221156 representing NM_022876 Red=Cloning site Green=Tags(s) MAMSSGGSGGGVPEQEDSVLFRRGTGQSDDSDIWDDTALIKAYDKAVASFKHALKNGDICETSGKPKTTP KRKPAKKNKSQKKNTAASLQQWKVGDKCSAIWSEDGCIYPATIASIDFKRETCVVVYTGYGNREEQNLSD LLSPICEVANNIEQNAQENENESQVSTDESENSRSPGNKSDNIKPKSAPWNSFLPPPPPMPGPRLGPGKI IPPPPPICPDSLDDADALGSMLISWYMSGYHTGYYMGFRQNQKEGRCSHSLN TRTRPLEQKLISEEDLAANDILDYKDDDDKV
Tag:	C-Myc/DDK
Purity:	> 80% as determined by SDS-PAGE and Coomassie blue staining
Concentration:	>0.05 µg/µL as determined by microplate BCA method
Labeling Method:	Labeled with [U- 13C6, 15N4]-L-Arginine and [U- 13C6, 15N2]-L-Lysine
Buffer:	25 mM Tris-HCl, 100 mM glycine, pH 7.3
Storage:	Store at -80°C. Avoid repeated freeze-thaw cycles.
Stability:	Stable for 3 months from receipt of products under proper storage and handling conditions.
RefSeq:	NP_075014
RefSeq Size:	1527
RefSeq ORF:	786
Synonyms:	BCD541; C-BCD541; GEMIN1; SMNC; TDRD16B
Locus ID:	6607
UniProt ID:	Q16637

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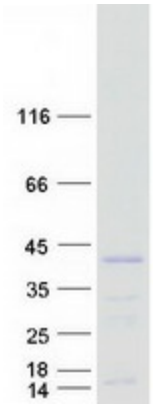
Cytogenetics: 5q13.2

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 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]).