

SMN EIA Kit

Sensitive Assay for Cell Lysates

Survival Motor Neuron (SMN) is a ~38 kDa protein produced chiefly by the SMN1 gene, located on the telomeric portion of chromosome 5q [1-4]. A nearly identical centromeric copy of the gene (SMN2) also produces a small amount of full-length SMN protein, but due to a translationally silent C(R)T transition that results in alternative splicing of the pre-mRNA, most of the resulting SMN is truncated, causing reduced protein stability and lower overall SMN levels [5-6]. Deletion or mutation of the SMN1 gene results in a reduced level of full-length SMN protein and manifests as a range of neuromuscular phenotypes in humans as the disease spinal muscular atrophy (SMA). SMA is characterized by muscle weakness and atrophy, functional disability and is the most common lethal genetic disease of infants and toddlers. Approximately one in 35 adults is a carrier of the SMN1 mutation. The incidence of SMA is 1 in 6,000 to 1 in 10,000 live births [7]. SMN protein is present in the cell cytoplasm, and also in the nucleus where it is concentrated in "gem" structures associated with Cajal bodies [8-9]. SMN protein is a constituent of Gemin-containing complexes, and is thought to participate in many aspects of RNA metabolism. SMN complexes have been shown to mediate the assembly of

uridine-rich small nuclear ribonucleoproteins (snRNPs), which in turn act as critical components of spliceosomes [10].

LIT: [1] Molecular mechanisms of spinal muscular atrophy: C. J. Sumner; *J. Child Neurol.* **22**, 979 (2007) • **[2]** Genetic mapping of chronic childhood-onset spinal muscular atrophy to chromosome 5q11.2-13.3: L. M. Brzustowicz, et al.; *Nature* **344**, 540 (1990) • **[3]** Mapping of acute (type I) spinal muscular atrophy to chromosome 5q12-q14. The French Spinal Muscular Atrophy Investigators: J. Melki, et al.; *Lancet* **336**, 271 (1990) • **[4]** Identification and characterization of a spinal muscular atrophy-determining gene: S. Lefebvre, et al.; *Cell* **80**, 155 (1995) • **[5]** SMN oligomerization defect correlates with spinal muscular atrophy severity: C. Lorson, et al.; *Nat. Genet.* **19**, 63 (1998) • **[6]** Regulation of SMN protein stability: B. G. Burnett, et al.; *Mol. Cell. Biol.* **29**, 1107 (2009) • **[7]** Incidence, prevalence, and gene frequency studies of chronic childhood spinal muscular atrophy: J. Pearn; *J. Med. Genet.* **15**, 409 (1978) • **[8]** The spinal muscular atrophy disease gene product, SMN, and its associated protein SIP1 are in a complex with spliceosomal snRNP proteins: Q. Liu, et al.; *Cell* **90**, 1013 (1997) • **[9]** The spinal muscular atrophy disease gene product, SMN: A link between snRNP biogenesis and the Cajal (coiled) body: T. Cavalho, et al.; *J. Cell Bio.* **147**, 715 (1999) • **[10]** The SMN-SIP1 complex has an essential role in spliceosomal snRNP biogenesis: U. Fischer, et al.; *Cell* **90**, 1023 (1997)

The SMN EIA kit was developed in collaboration with the SMA Foundation. Visit www.smafoundation.org for more information, and to share your research results with the SMA research community.

Spinal Muscular Atrophy
Foundation

SMN EIA Kit

NEW

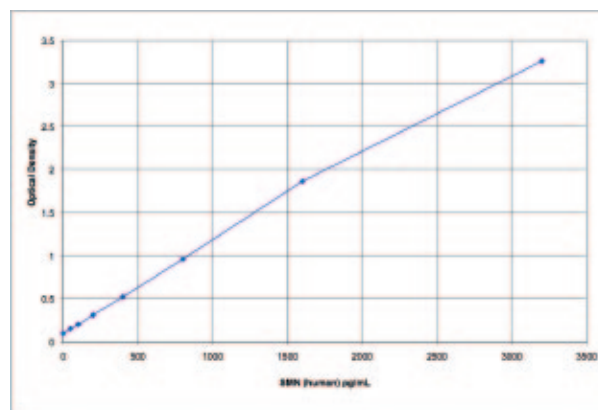
ADI-900-209

1 x 96 wells

APPLICATION: For the quantitative determination of human and mouse SMN in cell lysate samples. **SENSITIVITY:** 50 pg/ml (range 50-3200 pg/ml)

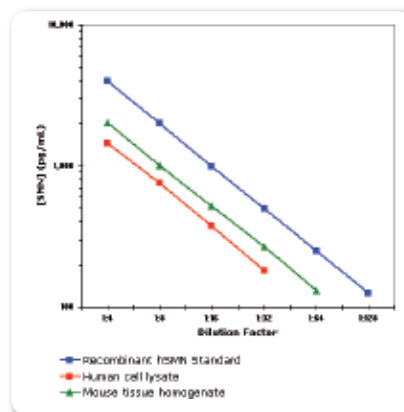
Product Features & Benefits

- **Sensitive**
Measure as little as 50 pg/ml of SMN
- **Quantitative**
Fully quantitative results surpass semi-quantitative Western blot analysis
- **Convenient**
Ready-to-use liquid color coded reagents and pre-coated 96-well plate
- **Time Saving**
Results from up to 39 samples in duplicate in just 3 hours



Parallelism Between Recombinant and Natural SMN

Parallelism experiments were carried out to determine if the recombinant human SMN standard accurately determines human and mouse SMN concentrations in biological matrices. To assess parallelism, values for human cell lysate and mouse tissue homogenate were obtained from a standard curve using four parameter logistic curve fitting. The observed concentration was plotted against the dilution factor. Parallelism of the curves demonstrates that the antibody binding characteristics are similar enough to allow the accurate determination of analyte levels in diluted samples.



Related Protein

Product	Source/Host	Prod. No.	Size
SMN1 (human), (rec.) (His-tag)	Produced in <i>E. coli</i> . Human SMN1 is fused at N-terminus with a His tag.	ADI-NBP-201-050	50 µg

SMN and Chaperones – Small Heat Shock Proteins, SMN and Inherited Motor Neuron Disease

Similar to the etiology of spinal muscular atrophy, mutations in the genes encoding the small heat shock proteins Hsp22 (HspB8) and Hsp27 (HspB1) have been associated with the inherited motor neuron diseases (MND) distal hereditary motor neuropathy and Charcot-Marie-Tooth disease, respectively. Recent findings by Sun et al. suggest SMN, HspB8, and HspB1 may act through a common mechanism involving the DEAD box protein Ddx20, where mutation in these genes disrupts spliceosome assembly and pre-mRNA processing.

LIT: Abnormal interaction of motor neuropathy-associated mutant HspB8 (Hsp22) forms with the RNA helicase Ddx20 (gemin3): X. Sun, et al.; Cell Stress Chaperones **15**, 567 (2010)

Related Products

Product	Prod. No.
HSP27 (human), EIA kit	ADI-EKS-500
ImmunoSet™ HSP27 high sensitivity (human), ELISA development set	ADI-960-076
HSP27 (human), (rec.)	ADI-SPP-715
HSP27, mAb (G3.1)	ADI-SPA-800
HSP27, mAb (G3.1) (FITC conjugate)	ADI-SPA-800FI
HSP27, pAb	ADI-SPA-803
[pSer¹⁵]HSP27, pAb	ADI-SPA-525
[pSer⁷⁸]HSP27, pAb	ADI-SPA-523
[pSer⁸²]HSP27, pAb	ADI-SPA-524
ImmunoSet™ HSP25 (rodent), ELISA development set	ADI-960-075
HSP25, pAb	ADI-SPA-801

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