

SMN (h2): 293T Lysate: sc-113432

BACKGROUND

Spinal muscular atrophy (SMA) is an autosomal recessive neurodegenerative disease characterized by loss of motor neurons in the spinal cord. SMA is caused by deletion or loss-of-function mutations of SMN (survival of motor neuron) gene. SMN, also known as Gemin1, SMN1, SMNT and BCD541, exists as four isoforms produced by alternative splicing. SMN is oligomeric and forms a complex with Gemin2 (formerly SIP1), Gemin3 (a DEAD box RNA helicase), Gemin4, Gemin5 and Gemin6, as well as several spliceosomal snRNP proteins. The SMN complex plays an essential role in spliceosomal snRNP assembly in the cytoplasm and is required for pre-mRNA splicing of the nucleus. The SMN complex is found in both the cytoplasm and the nucleus. The nuclear form is concentrated in subnuclear bodies called gems (gemini of the coiled bodies). Cytoplasmic SMN interacts with spliceosomal Sm proteins and facilitates their assembly onto U snRNAs, and nuclear SMN mediates recycling of pre-mRNA splicing factors. Nearly identical telomeric and centromeric forms of SMN encode the same protein; however, only mutations in the telomeric form are associated with the disease-state SMA. SMN is expressed in a wide variety of tissues including brain, kidney, liver, spinal cord and moderately in skeletal and cardiac muscle.

REFERENCES

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- Fischer, U., et al. 1997. The SMN-SIP1 complex has an essential role in spliceosomal snRNP biogenesis. *Cell* 90: 1023-1029.
- Monani, U., et al. 1999. A single nucleotide difference that alters splicing patterns distinguishes the SMA gene SMN1 from the copy gene SMN2. *Hum. Mol. Genet.* 8: 1177-1183.
- Meister, G., et al. 2000. Characterization of a nuclear 20S complex containing the survival of motor neurons (SMN) protein and a specific subset of spliceosomal Sm proteins. *Hum. Mol. Genet.* 9: 1977-1986.
- Mourelatos, Z., et al. 2001. SMN interacts with a novel family of hnRNP and spliceosomal proteins. *EMBO J.* 20: 5443-5452.
- Online Mendelian Inheritance in Man, OMIM™. 2008. Johns Hopkins University, Baltimore, MD. MIM Number: 600354. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
- LocusLink Report (LocusID: 6606). <http://www.ncbi.nlm.nih.gov/LocusLink/>

CHROMOSOMAL LOCATION

Genetic locus: SMN1 (human) mapping to 5q13.2.

PRODUCT

SMN (h2): 293T Lysate represents a lysate of human SMN transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

APPLICATIONS

SMN (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive SMN antibodies.

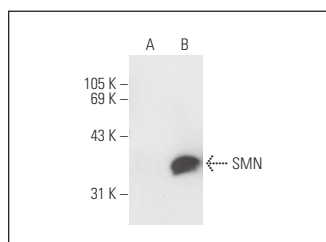
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

SMN (H-7): sc-137200 is recommended as a positive control antibody for Western Blot analysis of enhanced human SMN expression in SMN transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

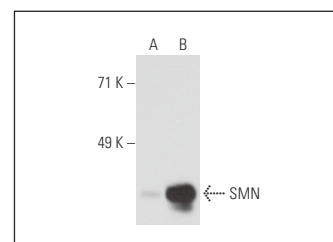
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended:
1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA



SMN (H-7): sc-137200. Western blot analysis of SMN expression in non-transfected: sc-117752 (A) and human SMN transfected: sc-113432 (B) 293T whole cell lysates.



SMN (SCB3): sc-101365. Western blot analysis of SMN expression in non-transfected: sc-117752 (A) and human SMN transfected: sc-113432 (B) 293T whole cell lysates.

RESEARCH USE

For research use only, not for use in diagnostic procedures.

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.