

## Produktinformation



Forschungsprodukte & Biochemikalien
Zellkultur & Verbrauchsmaterial
Diagnostik & molekulare Diagnostik
Laborgeräte & Service

Weitere Information auf den folgenden Seiten! See the following pages for more information!



Lieferung & Zahlungsart siehe unsere Liefer- und Versandbedingungen

## Zuschläge

- Mindermengenzuschlag
- Trockeneiszuschlag
- Gefahrgutzuschlag
- Expressversand

## SZABO-SCANDIC HandelsgmbH

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SMN2 (Huma	n) IP-WB Antibody Pair	
atalog # : H0000	06607-PW2 規格:[1 Set]	
ist All		
Specification		Application Image
Product Description:	This IP-WB antibody pair set comes with one antibody for immunoprecipitation and another to detect the precipitated protein in western blot.	Immunoprecipitation-Western Blot
Reactivity:	Human	
Quality Control Testing:	Immunoprecipitation-Western Blot (IP-WB)	
Supplied Product:	Antibody pair set content: 1. Antibody pair for IP: rabbit polyclonal anti-SMN2 (300 ul) 2. Antibody pair for WB: mouse purified polyclonal anti-SMN2 (50 ug)	
Storage Instruction:	Store reagents of the antibody pair set at -20°C or lower. Please aliquot to avoid repeated freeze thaw cycle. Reagents should be returned to -20°C storage immediately after use.	
Applications		
Immunoprecipit	tation-Western Blot	
Gene Informatio	on	
Entrez GenelD:	<u>6607</u>	
Gene Name:	SMN2	
Gene Alias:	BCD541,C-BCD541,FLJ76644,MGC20996,MGC5208,SMNC	
Gene Description:	survival of motor neuron 2, centromeric	
Omim ID:	<u>601627</u>	
Gene Ontology	: <u>Hyperlink</u>	
0		

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

Other OTTHUMP00000125236,OTTHUMP00000125237,gemin 1
Designations:

## **Related Disease**

Amyotrophic Lateral Sclerosis Amyotrophic lateral sclerosis Disease Progression Genetic Predisposition to Disease Muscular Atrophy, Spinal Nerve Degeneration Spinal Muscular Atrophies of Childhood Spinal muscular atrophy

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