

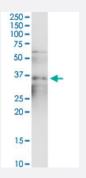
#### MaxPab®

# SMN2 MaxPab rabbit polyclonal antibody (D01)

Catalog # H00006607-D01

Size 100 uL

## Applications



### Immunoprecipitation

Immunoprecipitation of SMN2 transfected lysate using anti-SMN2 MaxPab rabbit polyclonal antibody and Protein A Magnetic Bead, and immunoblotted with SMN2 purified MaxPab mouse polyclonal antibody (B01P) (H00006607-B01P).

Specification	
Product Description	Rabbit polyclonal antibody raised against a full-length human SMN2 protein.
Immunogen	SMN2 (NP_075013.1, 1 a.a. ~ 282 a.a) full-length human protein.
Sequence	MAMSSGGSGGGVPEQEDSVLFRRGTGQSDDSDIWDDTALIKAYDKAVASFKHALKNGDICETSG KPKTTPKRKPAKKNKSQKKNTAASLQQWKVGDKCSAIWSEDGCIYPATIASIDFKRETCVVVYTG YGNREEQNLSDLLSPICEVANNIEQNAQENENESQVSTDESENSRSPGNKSDNIKPKSAPWNSFL PPPPPMPGPRLGPGKPGLKFNGPPPPPPPPPPPHLLSCWLPPFPSGPPIIPPPPPICPDSLDDAD ALGSMLISWYMSGYHTGYYMEMLA
Host	Rabbit
Reactivity	Human
Quality Control Testing	Antibody reactive against mammalian transfected lysate.
Storage Buffer	No additive
Storage Instruction	Store at -20°C or lower. Aliquot to avoid repeated freezing and thawing.



### Applications

Immunoprecipitation

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Protocol Download

### Gene Info — SMN2

Entrez GenelD	<u>6607</u>
GeneBank Accession#	<u>NM_022875</u>
Protein Accession#	<u>NP_075013.1</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	Hyperlink
Gene Summary	This gene is part of a 500 kb inverted duplication on chromosome 5q13. This duplicated region c ontains at least four genes and repetitive elements which make it prone to rearrangements and d eletions. The repetitiveness and complexity of the sequence have also caused difficulty in determi ning the organization of this genomic region. The telomeric and centromeric copies of this gene a re nearly identical and encode the same protein. While mutations in the telomeric copy are associ ated with spinal muscular atrophy, mutations in this gene, the centromeric copy, do not lead to dis ease. This gene may be a modifier of disease caused by mutation in the telomeric copy. The criti cal sequence difference between the two genes is a single nucleotide in exon 7, which is thought t o be an exon splice enhancer. Note that the nine exons of both the telomeric and centromeric copi es 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 e ncoded by this gene localizes to both the cytoplasm and the nucleus. Within the nucleus, the protei n 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 invo lved 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 Ref Seq
Other Designations	OTTHUMP00000125236 OTTHUMP00000125237 gemin 1



#### Disease

- <u>Amyotrophic lateral sclerosis</u>
- Disease Progression
- Genetic Predisposition to Disease
- <u>Muscular Atrophy</u>
- <u>Nerve Degeneration</u>
- Spinal Muscular Atrophies of Childhood
- Spinal muscular atrophy