DNAxPAb | Hard-to-Find Antibody

SMN2 DNAxPab

Catalog # H00006607-W02P Size 200 ug

Specification

Product Description	Rabbit polyclonal antibody raised against a full-length human SMN2 DNA using DNAx™ Immune tec hnology.
Technology	DNAx™ Immune
Immunogen	Full-length human DNA
Sequence	MAMSSGGSGGGVPEQEDSVLFRRGTGQSDDSDIWDDTALIKAYDKAVASFKHALKNGDICETSG KPKTTPKRKPAKKNKSQKKNTAASLQQWKVGDKCSAIWSEDGCIYPATIASIDFKRETCVVVYTG YGNREEQNLSDLLSPICEVANNIEQNAQENENESQVSTDESENSRSPGNKSDNIKPKSAPWNSFL PPPPPMPGPRLGPGKPGLKFNGPPPPPPPPPHLLSCWLPPFPSGPPIIPPPPPICPDSLDDAD ALGSMLISWYMSGYHTGYYMGFRQNQKEGRCSHSLN
Host	Rabbit
Reactivity	Human
Purification	Protein A
Quality Control Testing	Antibody reactive against mammalian transfected lysate.
Storage Buffer	In 1x PBS, pH 7.4
Storage Instruction	Store at -20°C or lower. Aliquot to avoid repeated freezing and thawing.

Applications

- Western Blot (Transfected lysate)

  Protocol Download

- Immunofluorescence (Transfected cell)
- Flow Cytometry (Transfected cell)

Gene Info — SMN2

Entrez GeneID	6607
GeneBank Accession#	DQ894734.2
Protein Accession#	ABM85660.1
Gene Name	SMN2
Gene Alias	BCD541, C-BCD541, FLJ76644, MGC20996, MGC5208, SMNC
Gene Description	survival of motor neuron 2, centromeric
Omim ID	601627
Gene Ontology	Hyperlink
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 Designations	OTTHUMP00000125236|OTTHUMP00000125237|gemin 1

Disease

- Amyotrophic lateral sclerosis
- Disease Progression
- Genetic Predisposition to Disease

- Muscular Atrophy
- Nerve Degeneration
- Spinal Muscular Atrophies of Childhood
- Spinal muscular atrophy