10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

SMUBP2 Polyclonal Antibody

Purified Rabbit Polyclonal Antibody (Pab)
Catalog # AP54619

Product Information

Application	WB, IHC-P, IHC-F, IF, ICC, E
Primary Accession	P38935
Reactivity	Rat, Pig, Bovine
Host	Rabbit
Clonality	Polyclonal
Calculated MW	109149
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human SMUBP2
Epitope Specificity	271-355/993
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Nucleus. Cytoplasm.
SIMILARITY	Belongs to the DNA2/NAM7 helicase family. Contains 1 AN1-type zinc finger. Contains 1 R3H domain.
SUBUNIT	Homooligomer. Interacts with RUVBL1, RUVBL2, GTF3C1 andABT1. Is part of large cytosolic ribonucleoprotein complexes(Probable). Associates with the ribosomes.
Post-translational modifications	Phosphorylated upon DNA damage, probably by ATM or ATR.
DISEASE	Defects in IGHMBP2 are the cause of distal hereditary motor neuronopathy type 6 (HMN6) [MIM:604320]; also known as spinal muscular atrophy distal autosomal recessive 1 (DSMA1) or spinal muscular atrophy with respiratory distress 1 (SMARD1). Distal hereditary motor neuronopathies constitute a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. The most prominent symptoms of HMN6 are severe respiratory distress resulting from diaphragmatic paralysis with eventration shown on chest x-ray and predominant involvement of the upper limbs and distal muscles.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	IGHMBP2 is a 993 amino acid nuclear and cytoplasmic protein that is ubiquitously expressed. Belonging to the DNA2/NAM7 helicase family, IGHMBP2 is a 5' to 3' helicase that unwinds RNA and DNA duplexes in an ATP-dependent reaction. IGHMBP2 also acts as a transcriptional regulator and is necessary for transcriptional activation of the flounder liver-type antifreeze protein gene. IGHMBP2 exists as a homooligomer and is part of the cytosolic

ribonucleoprotein complex. Mutations in the gene encoding IGHMBP2 are suggested to lead to distal hereditary motor neuronopathy type 6 (HMN6), also known as spinal muscular atrophy distal autosomal recessive 1 (DSMA1) or spinal muscular atrophy with respiratory distress 1 (SMARD1). HMN6 is characterized by weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs and severe respiratory distress.

Additional Information

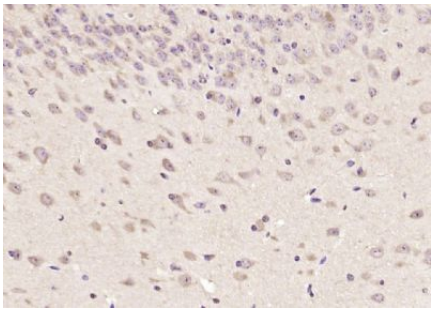
Gene ID	3508
Other Names	DNA-binding protein SMUBP-2, 3.6.4.12, 3.6.4.13, ATP-dependent helicase IGHMBP2, Glial factor 1, GF-1, Immunoglobulin mu-binding protein 2, IGHMBP2, SMBP2, SMUBP2
Target/Specificity	Expressed in all tissues examined.
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC=1:100-500,IF=1:100-500,ELISA=1:5000-10000
Format	0.01M TBS(pH7.4) with 1% BSA, 0.09% (W/V) sodium azide and 50% Glyce
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Name	IGHMBP2
Synonyms	SMBP2, SMUBP2
Function	5' to 3' helicase that unwinds RNA and DNA duplexes in an ATP-dependent reaction (PubMed:19158098, PubMed:22999958, PubMed:30218034). Specific to 5'-phosphorylated single-stranded guanine-rich sequences (PubMed:22999958, PubMed:8349627). May play a role in RNA metabolism, ribosome biogenesis or initiation of translation (PubMed:19158098, PubMed:19299493). May play a role in regulation of transcription (By similarity). Interacts with tRNA-Tyr (PubMed:19299493).
Cellular Location	Nucleus. Cytoplasm. Cell projection, axon {ECO:0000250|UniProtKB:P40694}
Tissue Location	Expressed in all tissues examined. Expressed in the developing and adult human brain, with highest expression in the cerebellum. Moderately expressed in fibroblasts

Images

Paraformaldehyde-fixed, paraffin embedded (mouse brain); Antigen retrieval by boiling in sodium citrate buffer (pH6.0) for 15min; Block endogenous peroxidase by 3% hydrogen peroxide for 20 minutes; Blocking buffer (normal goat serum) at 37°C for 30min; Antibody incubation with (SMUBP2) Polyclonal Antibody,



Unconjugated (AP54619) at 1:200 overnight at 4°C, followed by operating according to SP Kit(Rabbit) (sp-0023) instructionsand DAB staining.