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

Senataxin Rabbit pAb

Senataxin Rabbit pAb
Catalog # AP54598

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q7Z333
Reactivity	Mouse
Predicted	Rat, Dog, Human, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	302880
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human Senataxin
Epitope Specificity	321-398/2677
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	nucleolus. Cytoplasm. May be detected in the nucleolus only in cycling cells (By similarity). Most abundant in the nucleus. Detected in granules. Colocalized in cycling cells with FBL in the nucleolus.
DISEASE	Defects in SETX are the cause of spinocerebellar ataxia autosomal recessive type 1 (SCAR1) [MIM:606002]; also known as ataxia-ocular apraxia 2. Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCAR1 is an autosomal recessive form associated with peripheral neuropathy and elevated serum alpha-fetoprotein, immunoglobulins and, less commonly, creatine kinase levels. Some SCAR1 patients manifest oculomotor apraxia. Defects in SETX are a cause of amyotrophic lateral sclerosis type 4 (ALS4) [MIM:602433]. ALS4 is a familial form of amyotrophic lateral sclerosis, a neurodegenerative disorder affecting upper and lower motor neurons and resulting in fatal paralysis. Sensory abnormalities are absent. Death usually occurs within 2 to 5 years. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of cases leading to familial forms. ALS4 is a childhood- or adolescent-onset form characterized by slow disease progression and the sparing of bulbar and respiratory muscles.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	SETX belongs to the DNA2/NAM7 helicase family. Localizing to the nucleolus or the nucleoplasm in a cell cycle-dependent manner and to the cytoplasm, SETX contains a C-terminal DNA/RNA helicase domain and is believed to function as a helicase involved in RNA processing and DNA repair. Mutations in the gene encoding SETX can lead to ataxia-ocular apraxia 2 (AOA2) or amyotrophic lateral sclerosis 4 (ALS4). AOA2, also known as spinocerebellar

ataxia-1 (SCAR1), is an autosomal recessive disorder characterized by progressive neurodegeneration of the cerebellum associated with the loss of Purkinje cells. ALS4 is a familial childhood- or adolescent-onset neurodegenerative disorder affecting both upper and lower motor neurons that ultimately results in fatal paralysis.

Additional Information

Gene ID 23064

Other Names Helicase senataxin, 3.6.4.12, 3.6.4.13, Amyotrophic lateral sclerosis 4 protein, SEN1 homolog, Senataxin {ECO:0000303|PubMed:14770181, ECO:0000312|HGNC:HGNC:445}, SETX {ECO:0000303|PubMed:14770181, ECO:0000312|HGNC:HGNC:445}

Dilution IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500

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 SETX {ECO:0000303|PubMed:14770181, ECO:0000312|HGNC:HGNC:445}

Function ATP-dependent 5'-3' helicase that preferentially unwinds RNA:DNA substrates over DNA:DNA substrates, playing a crucial role in resolving 5'-overhang RNA:DNA hybrids (R-loops) and promoting transcription termination (PubMed:36864660). Plays a role in transcription regulation by its ability to modulate RNA Polymerase II (Pol II) binding to chromatin and through its interaction with proteins involved in transcription (PubMed:19515850, PubMed:21700224). Contributes to the mRNA splicing efficiency and splice site selection (PubMed:19515850). Required for the resolution of R-loop RNA-DNA hybrid formation at G-rich pause sites located downstream of the poly(A) site, allowing XRN2 recruitment and XRN2-mediated degradation of the downstream cleaved RNA and hence efficient RNA polymerase II (RNAp II) transcription termination (PubMed:19515850, PubMed:21700224, PubMed:26700805). Required for the 3' transcriptional termination of PER1 and CRY2, thus playing an important role in the circadian rhythm regulation (By similarity). Involved in DNA double-strand breaks damage response generated by oxidative stress (PubMed:17562789). In association with RRP45, targets the RNA exosome complex to sites of transcription-induced DNA damage (PubMed:24105744). Plays a role in the development and maturation of germ cells: essential for male meiosis, acting at the interface of transcription and meiotic recombination, and in the process of gene silencing during meiotic sex chromosome inactivation (MSCI) (By similarity). May be involved in telomeric stability through the regulation of telomere repeat-containing RNA (TERRA) transcription (PubMed:21112256). Plays a role in neurite outgrowth in hippocampal cells through FGF8-activated signaling pathways. Inhibits retinoic acid-induced apoptosis (PubMed:21576111).

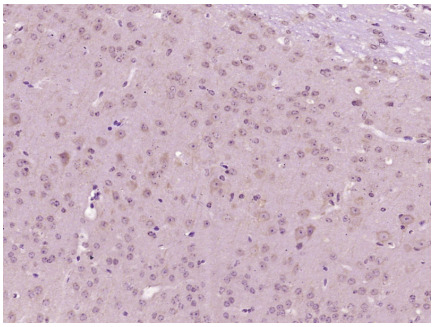
Cellular Location Nucleus. Nucleus, nucleoplasm. Nucleus, nucleolus. Cytoplasm. Chromosome Chromosome, telomere. Cell projection, axon. Cell projection, growth cone. Note=May be detected in the nucleolus only in cycling cells. At pachytene stage, colocalizes predominantly to the heterochromatic XY-body of sex

	chromosomes with DNA damage response proteins in a BRCA1-dependent manner (By similarity). Localizes with telomeric DNA in a transcription-dependent manner (PubMed:21112256) Under replication stress, colocalizes with a variety of DNA damage signaling and repair response proteins at distinct nuclear foci in mitotic S/G2- and G1-phase cells in a transcription- and RNA/DNA hybrid-dependent manner (PubMed:23149945). Localizes at limited number of nuclear foci (PubMed:24105744). Colocalizes with EXOSC9 in nuclear foci upon induction of transcription-related DNA damage at the S phase (PubMed:24105744). Most abundant in the nucleus. Detected in granules Colocalized in cycling cells with FBL in the nucleolus {ECO:0000250|UniProtKB:A2AKX3, ECO:0000269|PubMed:17562789, ECO:0000269|PubMed:21112256, ECO:0000269|PubMed:21576111, ECO:0000269|PubMed:23149945, ECO:0000269|PubMed:24105744}
Tissue Location	Highly expressed in skeletal muscle. Expressed in heart, fibroblast, placenta and liver. Weakly expressed in brain and lung. Expressed in the cortex of the kidney (highly expressed in tubular epithelial cells but low expression in the glomerulus)

Background

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

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 (Senataxin) Polyclonal Antibody, Unconjugated (AP54598) at 1:400 overnight at 4°C, followed by operating according to SP Kit(Rabbit) (sp-0023) instructionsand DAB staining.