

SmarcAL1 Rabbit pAb

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Catalog # AP57715

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q9NZC9
Predicted	Rat, Human, Mouse, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	105938
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human SmarcAL1
Epitope Specificity	581-680/954
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.
DISEASE	Defects in SMARCAL1 are a cause of Schimke immuno-osseous dysplasia (SIOD) [MIM:242900]. SIOD causes spondyloepiphyseal dysplasia, renal dysfunction and T-cell immunodeficiency. Approximately half of all patients also exhibit hyperthyroidism, while around half also exhibit episodal cerebral ischemia.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	The protein encoded by this gene is a member of the SWI/SNF family of proteins. Members of this family have helicase and ATPase activities and are thought to regulate transcription of certain genes by altering the chromatin structure around those genes. The encoded protein shows sequence similarity to the E. coli RNA polymerase-binding protein HepA. Mutations in this gene are a cause of Schimke immunoosseous dysplasia (SIOD), an autosomal recessive disorder with the diagnostic features of spondyloepiphyseal dysplasia, renal dysfunction, and T-cell immunodeficiency. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	50485
Other Names	SNF2 related chromatin remodeling annealing helicase 1, 3.6.4.-, 5.6.2.-, HepA-related protein, hHARP, SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A-like protein 1, Sucrose nonfermenting protein 2-like 1, SMARCAL1 (HGNC:11102)
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:500 0-10000

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.
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Protein Information

Name	SMARCAL1 (HGNC:11102)
Function	ATP-dependent annealing helicase that binds selectively to fork DNA relative to ssDNA or dsDNA and catalyzes the rewinding of the stably unwound DNA. Rewinds single-stranded DNA bubbles that are stably bound by replication protein A (RPA). Acts throughout the genome to reanneal stably unwound DNA, performing the opposite reaction of many enzymes, such as helicases and polymerases, that unwind DNA. May play an important role in DNA damage response by acting at stalled replication forks.
Cellular Location	Nucleus Note=Recruited to damaged DNA regions.
Tissue Location	Ubiquitously expressed, with high levels in testis.

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