

# Rabbit Anti-SmarcAL1 antibody

# SL19923R

Product Name:	SmarcAL1
Chinese Name:	蔗糖发酵蛋白2样蛋白1抗体
Alias:	HARP; HepA Related Protein; HepA-related protein; hHARP; SIOD; SMAL1_HUMAN; SMARCA like Protein 1; smarcal1; Sucrose nonfermenting protein 2 like 1; Sucrose nonfermenting protein 2-like 1; SWI/SNF Related; SWI/SNF related matrix associated actin dependent regulator of chromatin subfamily A like protein 1; SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A-like protein 1.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human, Mouse, Rat, Cow, Horse, Sheep,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	106kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human SmarcAL1:581-680/954
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20 °C. 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.
PubMed:	<u>PubMed</u>
Product Detail:	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]

#### Function:

ATP-dependent annealing helicase that 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.

#### **Subunit:**

Interacts with RPA2; the interaction is direct and mediates the recruitment by the RPA complex of SMARCAL1 to sites of DNA damage.

#### **Subcellular Location:**

Nucleus.

## Tissue Specificity:

Ubiquitously expressed, with high levels in testis.

#### **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 ischema.

#### Similarity:

Belongs to the SNF2/RAD54 helicase family.

SMARCAL1 subfamily.

Contains 2 HARP domains.

Contains 1 helicase ATP-binding domain.

Contains 1 helicase C-terminal domain.

#### **SWISS:**

Q9NZC9

#### Gene ID:

50485

#### Database links:

Entrez Gene: 338072 Cow

Entrez Gene: 50485 Human

Entrez Gene: 54380 Mouse

Entrez Gene: 316477 Rat

Omim: 606622 Human

SwissProt: Q9TTA5 Cow

SwissProt: Q9NZC9 Human

SwissProt: Q8BJL0 Mouse

SwissProt: B4F769 Rat

Unigene: 516674 Human

Unigene: 274232 Mouse

Unigene: 34679 Rat

### **Important Note:**

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