

# SmarcAL1 (h2): 293T Lysate: sc-115969

## BACKGROUND

SmarcAL1 (SWI/SNF-related matrix-associated Actin-dependent regulator of chromatin subfamily A-like protein 1), also known as HARP (HepA-related protein) or HHARP, is a 954 amino acid member of the SWI/SNF family of helicase and ATPase proteins. Localized to the nucleus, SmarcAL1 is a ubiquitously expressed protein that functions in ATP-dependent nucleosome-remodeling activities. SmarcAL1 contains one conserved C-terminal Snf2 domain, one helicase ATP-binding domain and two HARP (HepA-related) domains. Defects in the gene encoding SmarcAL1 are the cause of Schimke immuno-osseous dysplasia (SIOD), an autosomal recessive disorder characterized by renal dysfunction, spondyloepiphyseal dysplasia and T cell immunodeficiency.

## REFERENCES

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2. Boerkoel, C.F., et al. 2002. Mutant chromatin remodeling protein SmarcAL1 causes Schimke immuno-osseous dysplasia. *Nat. Genet.* 30: 215-220.
3. Lou, S., et al. 2002. Longevity in Schimke immuno-osseous dysplasia. *J. Med. Genet.* 39: 922-925.
4. Lücke, T., et al. 2005. Schimke-immuno-osseous dysplasia: new mutation with weak genotype-phenotype correlation in siblings. *Am. J. Med. Genet. A* 135A: 202-205.
5. Kilic, S.S., et al. 2005. Association of migraine-like headaches with Schimke immuno-osseous dysplasia. *Am. J. Med. Genet. A* 135A: 206-210.
6. Bökenkamp, A., et al. 2005. R561C missense mutation in the SmarcAL1 gene associated with mild Schimke immuno-osseous dysplasia. *Pediatr. Nephrol.* 20: 1724-1728.
7. Elizondo, L.I., et al. 2006. Schimke immuno-osseous dysplasia: a cell autonomous disorder? *Am. J. Med. Genet. A* 140A: 340-348.
8. Clewing, J.M., et al. 2007. Schimke immunoosseous dysplasia: suggestions of genetic diversity. *Hum. Mutat.* 28: 273-283.
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## CHROMOSOMAL LOCATION

Genetic locus: SMARCAL1 (human) mapping to 2q35.

## PRODUCT

SmarcAL1 (h2): 293T Lysate represents a lysate of human SmarcAL1 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

## STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

## APPLICATIONS

SmarcAL1 (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive SmarcAL1 antibodies. Recommended use: 10-20 µl per lane.

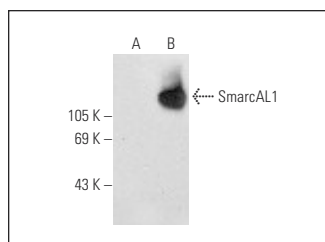
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

SmarcAL1 (E-12): sc-166209 is recommended as a positive control antibody for Western Blot analysis of enhanced human SmarcAL1 expression in SmarcAL1 transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

## RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

## DATA



SmarcAL1 (E-12): sc-166209. Western blot analysis of SmarcAL1 expression in non-transfected: sc-117752 (A) and human SmarcAL1 transfected: sc-115969 (B) 293T whole cell lysates.

## RESEARCH USE

For research use only, not for use in diagnostic procedures.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) for detailed protocols and support products.