

SRCAP gene

Snf2 related CREBBP activator protein

Normal Function

The *SRCAP* gene provides instructions for making a protein called Snf2-related CREBBP activator protein, or SRCAP. SRCAP is one of several proteins that help activate a gene called *CREBBP*. The protein produced from the *CREBBP* gene, called CREB binding protein, plays a key role in regulating cell growth and division and is important for normal development.

Health Conditions Related to Genetic Changes

Floating-Harbor syndrome

At least five *SRCAP* gene mutations have been identified in people with Floating-Harbor syndrome, a disorder involving short stature, slowing of the mineralization of the bones (delayed bone age), delayed speech development, and characteristic facial features.

The *SRCAP* gene mutations that cause Floating-Harbor syndrome may result in an altered protein that interferes with normal activation of the *CREBBP* gene, resulting in problems in development. However, the relationship between *SRCAP* gene mutations and the specific signs and symptoms of Floating-Harbor syndrome is unknown.

Other Names for This Gene

- domino homolog 2
- DOMO1
- EAF1
- FLHS
- helicase SRCAP
- KIAA0309
- Snf2-related CBP activator protein
- Snf2-related CREBBP activator protein
- SRCAP_HUMAN
- Swi2/Snf2-related ATPase homolog, domino homolog 1
- SWR1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SRCAP ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10847\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10847[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28SRCAP%5BTIAB%5D%29+OR+%28%28EAF1%5BTIAB%5D%29+OR+%28FLHS%5BTIAB%5D%29+OR+%28Snf2-related+CBP+activator+protein%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+engli sh%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- SNF2-RELATED CBP ACTIVATOR PROTEIN; SRCAP (<https://omim.org/entry/611421>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/10847>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SRCAP\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SRCAP[gene]))

References

- Hood RL, Lines MA, Nikkel SM, Schwartzentruber J, Beaulieu C, Nowaczyk MJ, Allanson J, Kim CA, Wieczorek D, Moilanen JS, Lacombe D, Gillesen-Kaesbach G, Whiteford ML, Quaio CR, Gomy I, Bertola DR, Albrecht B, Platzer K, McGillivray G, Zou R, McLeod DR, Chudley AE, Chodirker BN, Marcadier J; FORGE Canada Consortium; Majewski J, Bulman DE, White SM, Boycott KM. Mutations in SRCAP, encoding SNF2-related CREBBP activator protein, cause Floating-Harbor syndrome. *Am J Hum Genet.* 2012 Feb 10;90(2):308-13. doi: 10.1016/j.ajhg.2011.12.001. Epub 2012 Jan 19. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22265015>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3276662/>)
- Slupianek A, Yerrum S, Safadi FF, Monroy MA. The chromatin remodeling factor SRCAP modulates expression of prostate specific antigen and cellular proliferation in prostate cancer cells. *J Cell Physiol.* 2010 Aug;224(2):369-75. doi: 10.1002/jcp.22132. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20432434>)
- Wong MM, Cox LK, Chrivia JC. The chromatin remodeling protein, SRCAP, is critical for deposition of the histone variant H2A.Z at promoters. *J Biol Chem.* 2007

Sep 7;282(36):26132-9. doi: 10.1074/jbc.M703418200. Epub 2007 Jul 8. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17617668>)

Genomic Location

The *SRCAP* gene is found on chromosome 16 (<https://medlineplus.gov/genetics/chromosome/16/>).

Last updated December 1, 2012