



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

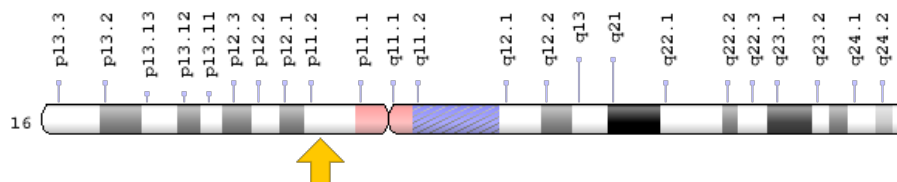
Floating-Harbor syndrome

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.

Chromosomal Location

Cytogenetic Location: 16p11.2, which is the short (p) arm of chromosome 16 at position 11.2

Molecular Location: base pairs 30,699,171 to 30,741,409 on chromosome 16 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Educational Resources

- Undiagnosed Diseases Network: Gene Page
<https://undiagnosed.hms.harvard.edu/genes/srcap/>

Clinical Information from GeneReviews

- Floating-Harbor Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK114458>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?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+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- SNF2-RELATED CBP ACTIVATOR PROTEIN
<http://omim.org/entry/611421>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SRCAP.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SRCAP%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:16974
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:10847>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/10847>
- UniProt
<https://www.uniprot.org/uniprot/Q6ZRS2>

Sources for This Summary

- Hood RL, Lines MA, Nikkel SM, Schwartzenruber J, Beaulieu C, Nowaczyk MJ, Allanson J, Kim CA, Wiczorek D, Moilanen JS, Lacombe D, Gillessen-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.
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- OMIM: SNF2-RELATED CBP ACTIVATOR PROTEIN
<http://omim.org/entry/611421>
- 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://www.ncbi.nlm.nih.gov/pubmed/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. Epub 2007 Jul 8.
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<https://ghr.nlm.nih.gov/gene/SRCAP>

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