

# Floating-Harbor Syndrome

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## SRCAP gene

Snf2 related CREBBP activator protein

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### 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.

More About This Health Condition

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

Scientific Articles on PubMed

PubMed

Catalog of Genes and Diseases from OMIM

SNF2-RELATED CBP ACTIVATOR PROTEIN; SRCAP

Gene and Variant Databases

NCBI Gene

ClinVar

## References

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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

**Genomic Location**The SRCAP gene is found on chromosome 16.

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