

SRCAP Gene

Subjects: Genetics

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Definition

Snf2 related CREBBP activator protein

1. 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.

2. Health Conditions Related to Genetic Changes

2.1. 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.

3. 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

References

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Keywords

genes

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