

Floating-Harbor Syndrome

Subjects: **Genetics & Heredity**

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Floating-Harbor syndrome is a disorder involving short stature, slowing of the mineralization of the bones (delayed bone age), delayed speech development, and characteristic facial features. The condition is named for the hospitals where it was first described, the Boston Floating Hospital and Harbor General Hospital in Torrance, California.

genetic conditions

1. Introduction

Growth deficiency in people with Floating-Harbor syndrome generally becomes apparent in the first year of life, and affected individuals are usually among the shortest 5 percent of their age group. Bone age is delayed in early childhood; for example, an affected 3-year-old child may have bones more typical of a child of 2. However, bone age is usually normal by age 6 to 12.

Delay in speech development (expressive language delay) may be severe in Floating-Harbor syndrome, and language impairment can lead to problems in verbal communication. Most affected individuals also have mild intellectual disability. Their development of motor skills, such as sitting and crawling, is similar to that of other children their age.

Typical facial features in people with Floating-Harbor syndrome include a triangular face; a low hairline; deep-set eyes; long eyelashes; a large, distinctive nose with a low-hanging separation (overhanging columella) between large nostrils; a shortened distance between the nose and upper lip (a short philtrum); and thin lips. As affected children grow and mature, the nose becomes more prominent.

Additionally some affected individuals have finger abnormalities that include short fingers (brachydactyly), widened and rounded tips of the fingers (clubbing), and curved pinky fingers (fifth finger clinodactyly). Other features of Floating-Harbor syndrome can include an unusually high-pitched voice and, in males, undescended testes (cryptorchidism).

2. Frequency

Floating-Harbor syndrome is a rare disorder; only about 50 cases have been reported in the medical literature.

3. Causes

Floating-Harbor syndrome is caused by mutations in the *SRCAP* gene. This 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 plays a key role in regulating cell growth and division and is important for normal development.

Mutations in the *SRCAP* gene 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. Rubinstein-Taybi syndrome, a disorder with similar features, is caused by mutations in the *CREBBP* gene itself.

3.1 The Gene Associated with Floating-Harbor Syndrome

- SRCAP

4. Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Most cases of Floating-Harbor syndrome result from new mutations in the gene and occur in people with no history of the disorder in their family. However, in some cases an affected person inherits the mutation from one affected parent.

5. Other Names for This Condition

- FHS
- FLHS
- Leisti-Hollander-Rimoin syndrome
- Pelletier-Leisti syndrome

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