

Floating-Harbor Syndrome

Subjects: **Genetics & Heredity**

Contributor: Nicole Yin

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

References

1. Arpin S, Afenjar A, Dubern B, Toutain A, Cabrol S, Héron D. Floating-Harbor Syndrome: report on a case in a mother and daughter, further evidence of autosomal dominant inheritance. *Clin Dysmorphol*. 2012 Jan;21(1):11-4. doi:10.1097/MCD.0b013e32834af5a7.
2. Asseidat I, Kaufman LM. Ocular abnormalities in Floating-Harbor syndrome. *JAAPOS*. 2009 Apr;13(2):218-20. doi: 10.1016/j.jaapos.2008.11.002.

3. Hood RL, Lines MA, Nikkel SM, Schwartzenbuber J, Beaulieu C, Nowaczyk MJ, Allanson J, Kim CA, Wieczorek D, Moilanen JS, Lacombe D, Gillessen-Kaesbach G, Whiteford ML, Quiao 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.
4. Lacombe D, Patton MA, Elleau C, Battin J. Floating-Harbor syndrome: description of a further patient, review of the literature, and suggestion of autosomal dominant inheritance. *Eur J Pediatr*. 1995 Aug;154(8):658-61. Review.
5. Nelson RA, McNamara M, Ellis W, Stein-Wexler R, Moghaddam B, Zwerdling T. Floating-Harbor syndrome and intramedullary spinal cord ganglioglioma: case report and observations from the literature. *Am J Med Genet A*. 2009 Oct;149A(10):2265-9. doi: 10.1002/ajmg.a.33014. Review.
6. Patton MA, Hurst J, Donnai D, McKeown CM, Cole T, Goodship J. Floating-Harbor syndrome. *J Med Genet*. 1991 Mar;28(3):201-4.
7. White SM, Morgan A, Da Costa A, Lacombe D, Knight SJ, Houlston R, Whiteford ML, Newbury-Ecob RA, Hurst JA. The phenotype of Floating-Harbor syndrome in 10 patients. *Am J Med Genet A*. 2010 Apr;152A(4):821-9. doi: 10.1002/ajmg.a.33294.

Retrieved from <https://www.encyclopedia.pub/entry/history/show/11445>