Floating-Harbor Syndrome

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Clinical characteristics:

Floating-Harbor syndrome (FHS) is characterized by typical craniofacial features; low birth weight, normal head circumference, and short stature; bone age delay that normalizes between ages six and 12 years; skeletal anomalies (brachydactyly, clubbing, clinodactyly, short thumbs, prominent joints, clavicular abnormalities); severe receptive and expressive language impairment; hypernasality and high-pitched voice; and intellectual disability that is typically mild to moderate. Difficulties with temperament and behavior that are present in many children tend to improve in adulthood. Other features can include hyperopia and/or strabismus, conductive hearing loss, seizures, gastroesophageal reflux, renal anomalies (e.g., hydronephrosis / renal pelviectasis, cysts, and/or agenesis), and genital anomalies (e.g., hypospadias and/or undescended testes).

Diagnosis/testing:

The diagnosis is established by identification of a heterozygous

pathogenic variant in those with clinical findings of FHS.

Management:

Treatment of manifestations:

Early intervention programs, special education, and vocational training to address developmental disabilities; communication rehabilitation with sign language or alternative means of communication; and behavior management by a behavioral specialist/psychologist with consideration of medication as needed. Referral to an endocrinologist for consideration of human growth hormone (HGH) therapy; however, data on use of HGH in FHS are limited. Standard treatment for refractive errors and strabismus, hearing loss, seizures, gastroesophageal reflux, and renal and genitourinary anomalies.

Surveillance:

Close monitoring of growth, especially in the first year. Annual: ophthalmologic evaluation, hearing

screening, blood pressure measurement, and assessment of renal function. Sonographic evaluation for renal cysts in teenage/adult years is indicated.

Genetic counseling:

FHS is inherited in an autosomal dominant manner. The majority of affected individuals have a de novo
pathogenic variant. Each child of an individual with FHS has a 50% chance of inheriting the pathogenic variant. Prenatal testing is possible for families in which the pathogenic variant has been identified.