CASE REPORT

Floating–Harbor Syndrome: A Rare Case Report

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Abstract

Aim and objective: To report a case with Floating–Harbor syndrome (FHS), emphasizing the general features and dental abnormalities and the treatment procedures and its outcome.

Background: FHS is an extremely rare genetic disorder, characterized by a triad: short stature, speech delay, and characteristic facies like triangular shape, bulbous nose, wide columella, deep-set eyes, long eyelashes, thin lips, short philtrum, and broad mouth. Approximately 50 cases have been described in the medical literature till date. Diagnosis is often delayed because the characteristic features of this syndrome are nonfamiliar.

Case description: A male child aged 5 years was referred to the dental OPD with the chief complaint of decayed upper and lower front and back teeth. On examination, the patient was found to have FHS along with the dental caries.

Conclusion: FHS is a rare genetic dysmorphic/mental retardation syndrome affecting both sexes but more among the female sex. There is no known cure for the disease and the treatment is symptomatic and supportive.

Clinical significance: An early diagnosis of FHS is important, as it enables with adequate information. These multiple malformations identification by an early diagnosis is crucial, as it requires a multidisciplinary approach in the initial evaluation, treatment, and follow-up.

Keywords: Clinodactly, Columella, Floating–Harbor syndrome, Microcephaly.

International Journal of Clinical Pediatric Dentistry (2020): 10.5005/jp-journals-10005-1816

Background

First described in 1973 by Pelletier and Feingold, Floating–Harbor syndrome (FHS) is an extremely rare genetic disorder. Also known as Pelletier-Leisti syndrome, this disease is named after the two hospitals, the Boston Floating Hospital and Harbor General Hospital in California, where the first cases were identified and reported by Robinson et al. (1974).[1] Till date, fewer than 50 cases of FHS have been described in the literature and is usually diagnosed in early childhood. FHS is characterized by a triad of delayed speech development, short stature, and characteristic facial appearance. Short neck, low birth weight, mild mental retardation, and brachydactyly are other less consistent features described in the literature. Some affected individuals exhibited additional features like finger abnormalities (fifth finger clinodactyly, brachydactyly, clubbing), hirsutism, low implanted ears with posterior rotation, celiac disease, voice with unusual high pitch, and undescended testes in males (cryptorchidism).[2] One of the major characteristics of this syndrome, delayed speech development, may be severe in some cases leading to language impairment and difficulties in verbal communication. Highly affected individuals have mild intellectual disability, though the motor skills are similar to the same-aged children. Although dental abnormalities are rare in this syndrome, malocclusion is the most noted abnormality and with rare occurrences of hypoplastic teeth, supernumerary teeth, agenesis of mandibular incisors, mandibular retrognathism, micrognathism, and hypoplastic jaw.[3] The present case report describes a patient with FHS, characteristics observed including the dental abnormalities, the treatment, and the result (Figs 1 and 2).

Case Description

- A male child aged 5 years was referred to the dental OPD with the chief complaint of decayed upper and lower front and back teeth. On examination, significant medical history was found.

  The child was born in the first pregnancy to nonconsanguineous phenotypically normal parents. At birth, parental age was 30 for mother and 33 for father. The mother had pregnancy preeclampsia and the pregnancy was completed by the lower segment cesarean section. Examination at birth revealed that the baby had perinatal asphyxia, meconium-stained amniotic fluid (MSAF), and he did not cry immediately after birth. Birth weight was 2.4 kg and height was 106 cm. The patient had persistent thrombocytopenia and was admitted in NICU for 11 days.
- After birth, he was prone to upper respiratory tract infections frequently. At 5 years of age when examined, it was noted that patient had microcephaly with alar facies, blue sclera, upslanting eyes, overhanging columella, prominent cup-shaped ears, reduced mouth opening, protruding tongue, triangular-shaped face with small chin, and fifth finger clinodactyly. He was identified with noticeable speech delay and behavioral inhibition, difficulty in adapting to new situations, and speaks loudly at times showing hyperactive behavior.
- Several investigations like 2D echo and DNA chromosome analysis, ophthalmology, ENT, and audiology evaluation were

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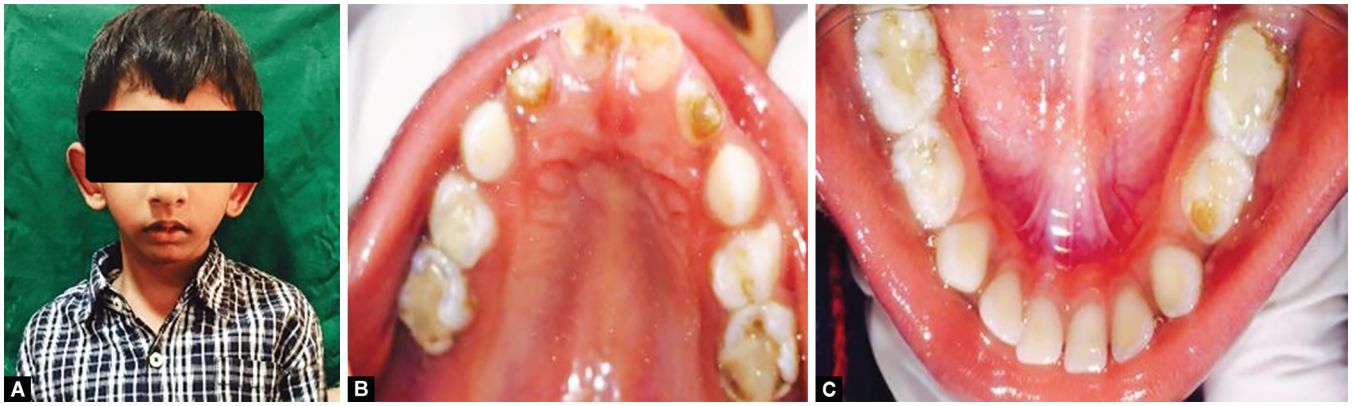
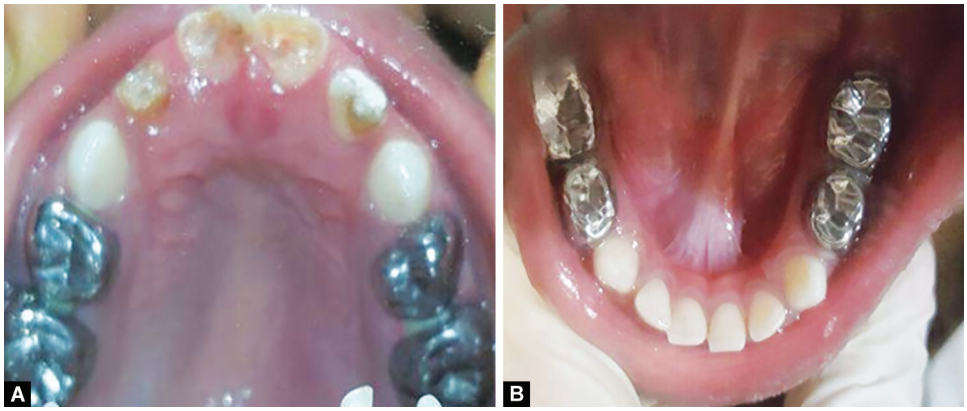
How to cite this article: Singana T, Suma NK, Sankriti AM. Floating–Harbor Syndrome: A Rare Case Report. Int J Clin Pediatr Dent 2020;13(5):569–571.

Source of support: Nil

Conflict of interest: None



Figs 1A to C: Preoperative photographs



Figs 2A and B: Postoperative photographs

carried out. In ENT evaluation, it was found that moderate hearing loss was seen with respect to the right and left ears. Ophthalmology evaluation was found normal. The 2D echo analysis has shown small atrioseptal defects.

- On intraoral examination, inflamed gingiva in relation to maxillary anteriors and normal appearing mucosa, freni, and tongue was observed. The patient had a full set of primary dentitions with mesial step molar relation on both right and left sides with anterior proclination and high arched palate. The patient had a habit of mouth breathing. The patient had undergone restorations in relation to 54, 55, 64, 65, 74, 75, 84, and 85. The mother reported the history of bottle feeding from 5 months to 4 years. 51, 52, 61, and 62 had deep dentinal caries suggestive of chronic irreversible pulpitis and secondary caries was noted in relation to 55, 64, 74, 84, and 85.
- The glass ionomer cement (GIC) restorations were done in relation to 55, 64, 74, and 84. Pulpectomy followed by composite restoration in relation to 52 was done; Pulpectomy followed by stainless steel crown in relation to 85 was performed. Stainless steel crowns in relation to 54, 55, 64, 65, 74, 75, and 84 were cemented. Extraction of 51, 61, and 62 was carried out.
- For the mouth breathing habit, underlying etiology, i.e., frequent upper respiratory tract infection, has to be corrected, which will allow this habit to correct automatically.

Discussion

The present case shows most of the features of FHS: triangular face, upslanting eyes, wide columella, low-set ears, short stature, and microcephaly. Other features like delay in speech development, intellectual disability, and poor social interaction were also noticed.[1–5]

Arpin et al.[4] in their study concluded that diagnosis of FHS is solely based on clinical findings as no laboratory tests are available and facial features being the most striking criteria.

This case report and the report published by De Benedetto et al.[1] showed no evidence of hirsutism or celiac disease, which sometimes has been associated with the syndrome. Cognitive abilities in FHS patients range from normal intelligence to mild mental retardation. In this patient, intellectual ability was subnormal, and he had learning difficulties that could be explained by language impairment. Delayed development of expressive speech was pointed out by all studies on FHS.

The third main feature of FHS is short stature. Almost all the reported cases had short stature.

This patient presented a mesial step in relation to both right and left sides with a high arched palate and poor oral hygiene with multiple carious lesions for which abnormal feeding practices might be the reason.

The clinical diagnosis of FHS may overlap with other dysmorphic syndromes such as Dubowitz syndrome, Rubinstein-Taybi syndrome (mutations in the CREBBP or EP300 genes), 3M syndrome, and Russell-Silver syndrome (abnormalities affecting certain genes on chromosomes 7 or 11). 3M syndrome and Russell-Silver syndrome are not associated with the expressive speech defect and this may prove to be a distinguishing feature of the diagnosis. Though the palatal abnormalities, broad nasal tip, and round face resemble Dubowitz syndrome in the infancy, it can be ruled out with absence of eczema and the developing facial features.

Conclusion

Floating–Harbor syndrome is a rare genetic dysmorphic/mental retardation syndrome affecting both sexes but more among the female sex. Most of the reported cases have sporadic occurrence but a few familial cases have been reported showing the possibility of the autosomal dominant mode of inheritance.

References

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