International Journal of Paediatric Dentistry 2004; **14:** 208–213

Floating–Harbor Syndrome: case report and craniofacial phenotype characterization

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Summary. Floating–Harbor syndrome is a rare genetic disorder of unknown aetiology. It was described for the first time in 1973. The syndrome is characterized mainly by short stature, delay in speech development and characteristic facial features. This article describes a report of a case of the syndrome and emphasizes the oral aspects, including descriptions of soft tissues, teeth, occlusion, stage of dental development and findings on examination of the temporomandibular joint. The treatment provided and its outcome is also described. Hopefully this information will be compared with findings from other patients in the future to assist in clarifying the phenotype of the Floating–Harbor syndrome.

Introduction

The Floating–Harbor syndrome is a rare genetic disorder which was described for the first time in 1973 by Pelletier & Feingold [1]. Its name was suggested by Leisti *et al*. [2] (1974), when they described the second case of the syndrome, and suggested the term derived from the names of the hospitals where they had been found – Boston Floating Hospital [1] and California's General Harbor Hospital [2].

Approximately 30 cases of Floating–Harbor syndrome have been described in medical literature up to the present. However, the true number may be less because the diagnosis in some of these cases was inconclusive [3,7–10,12,14–18]. The clinical features of the 25 clear cases of the Floating–Harbor syndrome described in the literature to date are summarized in Table 1. Floating–Harbor syndrome has been characterized by a triad of main features: short stature, speech delay and a characteristic facies, with triangular shape, bulbous nose, wide columella, deep-set eyes, long eyelashes, thin lips, short philtrum and broad mouth [1–18]. Other features have been described in the literature, such as low birth weight [1–5,9,10,14,15], short neck [1–4,6,9,11,12,16,17] brachydactyly and mild mental retardation [1–5,9,14–16,18], but have been less consistent. Features such as clinodactyly of the fifth finger [2–6,9–11,15–17], hirsutism [1,3,5,7,9,12,14,16,18] posteriorly rotated ears with low implantation [1–3,5,6,9,10,12,13,15,16–18] and celiac disease [3,6,9,12], have also been associated with the syndrome.

Reports of dental abnormalities in Floating–Harbor syndrome are rare. Malocclusion is the most cited and has been described in seven patients [2,3,5,7,16]. Some authors have commented on sporadically occurring anomalies, such as agenesis of mandibular incisors [13], supernumerary teeth [12], hypoplastic teeth [10], teeth malformation [5], mandibular retrognathism [14,16], micrognathism [2,16] and hypoplastic jaw [2,12].

The aim of this case report is to describe a patient with Floating–Harbor syndrome, emphasizing the general features and craniofacial characteristics, and to describe the treatment carried out and its outcome.

Case report

An 11-year-old boy sought assistance in the Group of Studies and Assistance of Disabled Patients of the Department of Pediatric Dentistry of the School of

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Table 1. Summary of abnormalities reported in patients with Floating–Harbor Syndrome.

Clinical manifestations	Previous cases	This case	Total
Consanguinity	1/16	–	1/17
Growth			
Low birth weight	10/23	+	11/24
Low birth length	9/15	*	9/15
Short stature	25/26	+	26/27
Delayed bone age	23/25	–	23/26
Development			
Delayed motor skills	7/25	+	7/26
Speech delay	27	+	28/28
Mild mental retardation	13/22	+	14/23
Craniofacial			
Triangular face	15/23	+	16/24
Bulbous nose	24/25	+	25/26
Wide columella	23/24	+	24/25
Short philtrum	22/25	+	23/26
Thin lips	23/24	+	24/25
Deep set eyes	15/22	+	17/23
Long eyelashes	21/24	+	22/25
Malocclusion	8/20	+	9/21
Posteriorly rotated ears	18/25	–	18/26
Other findings			
Hirsutism	12/21	–	12/22
Short neck	13/21	+	14/22
Clinodactyly	17/21	–	17/22
Brachydactyly	9/19	+	10/20
Clubbing	8/20	–	8/21
Joint laxity	8/18	+	9/19
Abdominal distension	7/17	*	7/17
Celiac disease	3/19	*	3/19
Strabismus	4/5	+	5/6

*not reported.

Dentistry – University of São Paulo. On presentation, the patient was examined by a physician; findings on general medical evaluation included: weight and height deficit, scarce subcutaneous tissue with well delineated muscles, heart murmur and interphalangeal joint laxity. Chromosomal examination showed a normal karyotype (46, XY). The anomalies found in the patient were considered to be suggestive of Floating–Harbor syndrome.

His parents were non-consanguineous; the mother was 36 and the father, 39 years old. The patient had been born through a natural birth after a full term pregnancy complicated only by emotional problems. The child had weighed 2·030 kg at birth and had been cyanotic.

At 9 days of age, the patient had a convulsion. He subsequently showed delay in development of early motor skills – he sat and walked after 2 years of age and had not started speaking until 9 years of age. At age 11, the patient showed learning difficulties; he attended a special school.

According to a speech and language evaluation, the patient presented a significant degree of articulatory disorder, his speech being nasal (velopharyngeal insufficiency) to an extent that impaired comprehension. He exhibited alterations to the position, tonicity and mobility of lips and tongue during speech. The parents reported significant improvement in communication after speech therapy.

The facial features of the patient included triangular face, short neck, bulbous nose, wide columella, short philtrum, deep-set eyes, long eyelashes, right eye meso-stabismus, broad mouth, thin lips (for someone of his age and ethnic group) and hypoplastic middle third of the face (Fig. 1). The soft tissue profile was frontally convex, the face appearing symmetrical and brachycephalic with competent lip seal. The clinical features of the patient described here together with those for patients described in previous reports are summarized in Table 1.

Temporomandibular joint (TMJ) evaluation was carried out but the patient showed no signs or symptoms of TMJ disorders. Maximum mouth opening was 45 mm with no midline shift during the opening. The mother reported that the child had shown bruxism up to the age of 6 years.

Analysis of his lateral cephalometric radiograph showed the patient to have retruded bone bases relative to the skull base and a convex skeletal profile. The pattern of the cephalic skeleton showed proportional growth. The upper and lower incisors were in protuded positions (Fig. 2).

On intraoral examination, the soft tissues showed no obvious changes, except for an upper labial frenum with low insertion. At initial examination, a large amount of dental plaque and gingivitis were evident. The patient showed a full permanent dentition, with low caries activity. The left primary mandibular lateral incisor was retained (or else this unit was a supplemental tooth), and there was a maxillary interincisal diastema caused by the upper labial frenum (Fig. 2). The patient showed a Class I malocclusion.

Although his chronological age was 11 years and 3 months, the carpal index for the child's radiographic examination indicated an age of 11 years and 6 months. This was analysed using the calcification stage of permanent teeth shown on panoramic radiography (Fig. 3). Compared with the stage of tooth development presented on the work of Nolla [19] (1960), the maxillary second premolars and third molars had a stage of calcification slightly in advance of average. Compared with the study of Nicodemo *et al*. [20]

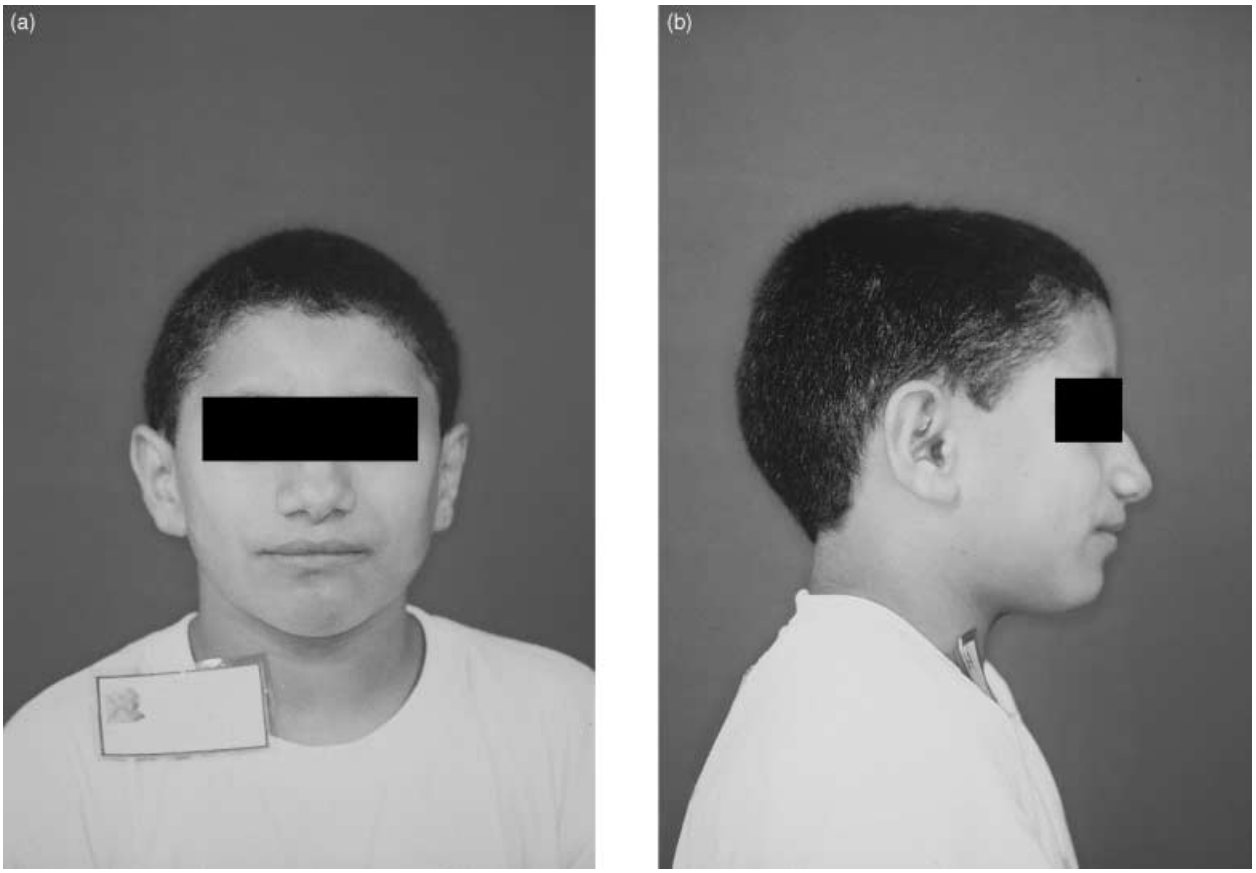

Fig. 1. Typical craniofacial appearance. (a) frontal view, (b) lateral view.

Table 2. Lateral cephalometric analysis.

Relationships	Mean	Cephalometric values	SD
F.NP	88°	86·8°	1·2°
NAP	0°	−3·1°	3·1°
SNA	82°	75·6°	6·4°
SNB	80°	75·3°	4·7°
ANB	2°	0·3°	1·7°
SND	76°	71·9°	4·1°
NS.Gn	67°	67·7°	0·7°
NS.PLO	14°	24·0°	10·0°
NS.GoGn	32°	37·1°	5·1°
GoGn.Plo	18°	13·1°	4·9°
1/./1	131°	116·5°	14·5°
1/.NS	103°	107·7°	4·7°
1/.NA	22°	32·2°	10·2°
1/–NA	4 mm	8·7 mm	4·7 mm
/1. NB	25°	31·1°	6·1°
/1-NB	4 mm	5·9 mm	1·9 mm
/1-NP	0 mm	3·7 mm	3·7 mm
1/–orbit	5 mm	4·6 mm	0·4 mm
/1-linha I	0 mm	−4·5 mm	4·5 mm
H. NB	9–12	9·0°	
H-nose	9–11	10·5 mm	
P-NB	0 mm	2·8 mm	2·8 mm
Mentom	8 mm	7·0 mm	1·0 mm

(1992), the canines, first and second premolars, seconds and third molars in the mandibular arch, and, first premolars, second, and third molars in the upper jaw presented a calcification stage which was also slightly advanced. Using Nolla's criteria, the majority of teeth were at a normal stage of calcification. Using the criteria of Nicodemo *et al*. [20] (1992), 50 per cent of teeth similarly presented a normal stage of development for age, whereas the other half were at a slightly more advanced stage of mineralization.

Conventional dental treatment was provided and included a maxillary frenectomy, extraction of the retained primary incisor and orthodontic treatment using both maxillary and mandibulary fixed appliances with no complications related to the patient's syndrome. Orthodontic treatment is currently still in progress (Fig. 4).

Discussion

The characteristic features described by other authors [1–7,9–13,15–18] – a triad of short stature, speech

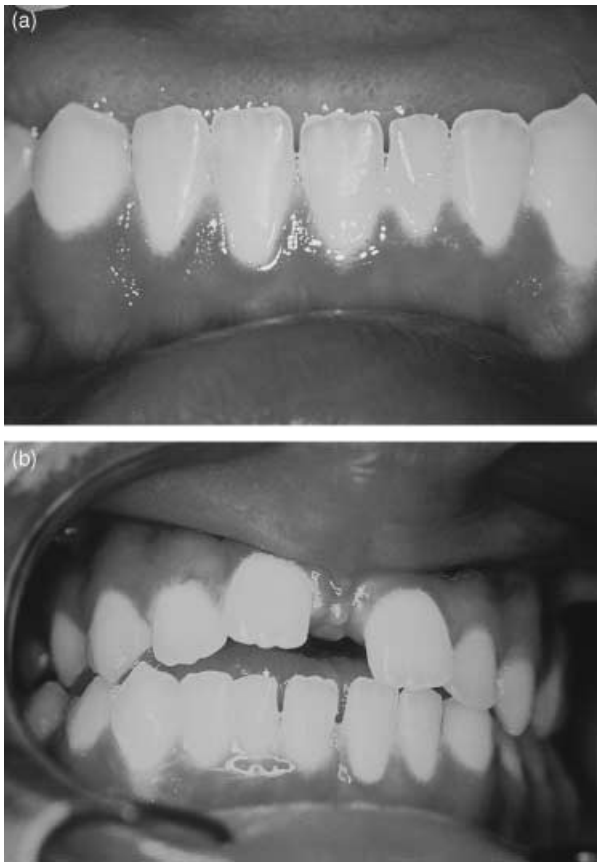

Fig. 2. Intraoral aspect. Note the diastema and Class I malocclusion.

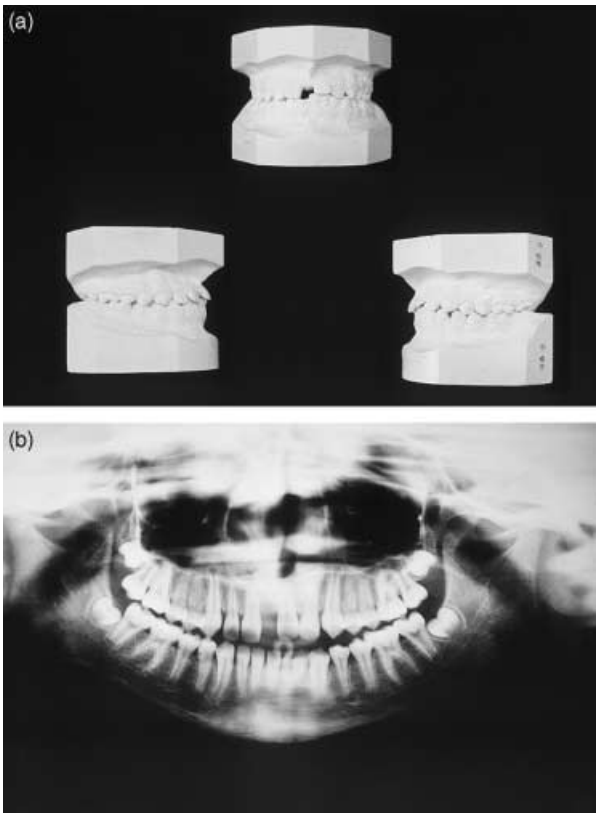

Fig. 3. Orthopantomograph showing the dentition at 11 years old.

delay and characteristic facies, with triangular shape, bulbous nose, wide columella, deep-set eyes, long eyelashes, thin lips, short philtrum and broad mouth were seen in this case. Other features described in the literature, such as low birth weight [1–5,9,10,14,15], short neck [1–4,6,9,11,12,16,17] brachydactyly and mild mental retardation [1–5,9,14–16,18] also presented in this case. There was however, no evidence of clinodactyly [2–6,9–11,15–17], hirsutism [1,3,5,7,9,12,14,16,18] or celiac disease [3,6,9,12], which have sometimes been associated with the syndrome.

Many authors have reported delay in bone age relative to chronological age [2–13,15–18], but in this case the patient presented carpal calcification compatible with his chronological age. In some previous reports, patients have been younger and perhaps this may be explained by the patient's age when the examination took place (11 years). Fryns *et al*. [14] (1993) also described a boy whose bone age corresponded to his chronological age at 16·5 years old. So, bone age delay may return to normal until it is compatible with chronological age. Wieczorek *et al*. (2001) [19] also related that delayed bone age may only be a charactheristic finding in young Floating–Harbor syndrome patients, but long-term follow-up is usually not reported in literature. These authors also reported a patient who received growth hormone therapy and subsequently showed normal height and near-normalization of bone age.

This patient presented a class I malocclusion with a median diastema between the upper permanent incisors, low insertion of the upper labial frenum and a retained left mandibular primary central incisor, in the presence of the erupted permanent successor.

The mineralization stage of the permanent teeth, verified through analysis using the criteria at both Nolla [20] (1960) and Nicodemo *et al*. [21] (1992) showed that second premolars and third upper molars were at a slightly advanced stage of mineralization. Considering the chronology proposed by Nicodemo *et al*. [21] (1992), 50 per cent of teeth had presented as being at the normal mineralization for age and 50 per cent were at a more advanced

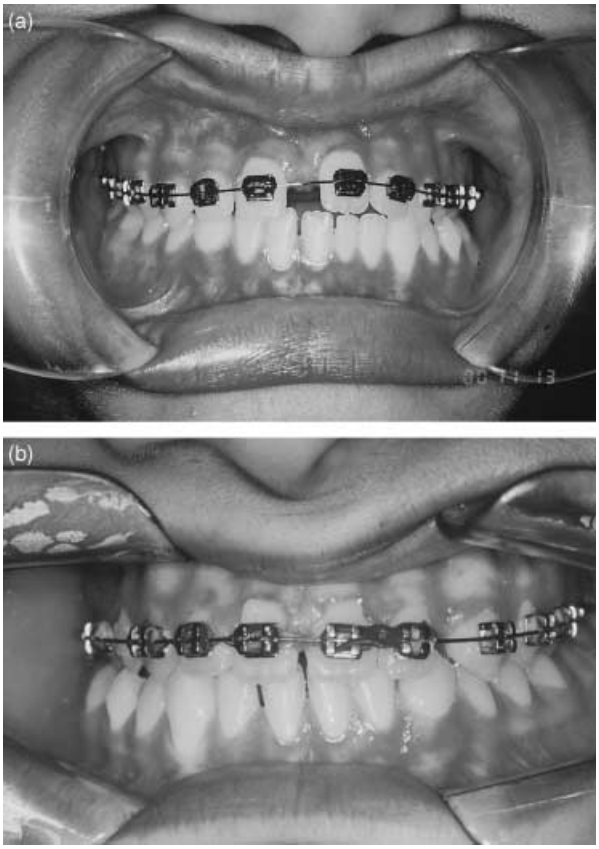

Fig. 4. Active orthodontic treatment at age 12 years.

stage of mineralization. The results of work by Nicodemo *et al*. [21] (1992) were obtained from Brazilian children, and this case is consistent with these results.

Although described in this article, it is not known whether the intraoral features are characteristic. These must be compared with future cases to clarify whether they are part of the phenotype of the syndrome or whether they are simply isolated signs.

Conclusion

In spite of the great number of authors who described the general medical features of patients with Floating–Harbor syndrome, there is a lack of information on intraoral findings in literature. Using the approach described in this article, other researchers may compare findings in future cases, so that the phenotype of the syndrome may be established. Dentists need to pay attention to the alterations related to the face of the patient, especially to features that may interfere with the normal development of the dentition of these patients as malocclusion seems to be a common aspect of the reports. Therefore, this aspect must be studied in these cases. In this case, treatment was carried out in the conventional way but this may not be true for all cases and the patient's systemic condition must be carefully evaluated.

Résumé. Le syndrome du Floating-Harbor est une maladie génétique rare d'origine inconnue. Il a été décrit pour la première fois en 1973. Ce syndrome est caractérisé par une petite taille, un retard de développement du langage et des caractéristiques faciales particulières. Cet article décrit un cas en insistant sur les aspects buccaux, notamment la description des tissus mous, des dents, de l'occlusion, du stade de développement dentaire et les données issues de l'examen de l'articulation temporo-mandibulaire. Le traitement et les résultats obtenus sont également décrits. Ces données devront, dans le futur, être comparées à celles issues d'autres patients, le phénotype de Floating-Harbor étant plus totalement établi.

Zusammenfassung. Das Floating-Harbor Syndrom ist eine seltene angeborene Erkrankung unbekannter Ätiologie. Es wurde erstmals 1973 beschrieben. Das Syndrom ist charakterisiert durch Minderwuchs, Sprachentwicklungsverzögerung und charakteristische Merkmale der Gesichtsmorphologie. Der vorliegende Artikel beschreibt einen Fall dieses Syndroms mit besonderer Berücksichtigung oraler Gewebe, einschließlich Beschreibung der Weichgewebe, Zähne, Okklusion, Entwicklungsstadium und Ergebnisse einer Untersuchung des Kiefergelenkes. Die Behandlung und das Therapieergebnis werden ebenfalls vorgestellt. Es ist anzustreben, zukünftig die Informationen mit den Daten anderer Patienten zu vergleichen, um so ein präziseres Bild des Phänotyps des Floating-Harbor Syndroms zu erhalten.

Resumen. El síndrome del Dique Flotante es una alteración genética rara de etiología desconocida. Se describió por primera vez en 1973. El síndrome se caracteriza principalmente por estatura corta, retraso en el desarrollo del habla y rasgos faciales característicos. Este artículo describe un caso y subraya los rasgos bucales, incluyendo descripciones de los tejidos blandos, dientes, oclusión, estadío del desarrollo dentario y los hallazgos del examen de

la articulación temporomandibular. También se describen el tratamiento aplicado y su resultado. Es deseable que en el futuro la información pueda compararse con la encontrada en otros pacientes y el fenotipo del Síndrome del Dique Flotante se establezca de forma más completa.

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