CASE REPORT

Frank–ter Haar syndrome—additional findings?

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Frank–ter Haar syndrome is a genetic disease that is transmitted by autosomal recessive pattern with characteristic features such as megalocornea or glaucoma, a prominent coccyx, heart defects, developmental delays, brachycephaly, a wide anterior fontanel, finger flexion deformities, full cheeks and micrognathia. Dentomaxillofacial features of this syndrome are not well documented in the literature. We present of a 21-year-old male with Frank–ter Haar syndrome and some features that may be linked with this syndrome not reported before in the literature.

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Introduction

Frank–ter Haar syndrome is a rare skeletal dysplasia with autosomal recessive inheritance that results from mutations in the *SH3PXD2B* gene.^{1,2} Frank–ter Haar syndrome is characterized by megalocornea or glaucoma, a prominent coccyx, heart defects, developmental delays, brachycephaly, a wide anterior fontanel, finger flexion deformities, full cheeks and micrognathia.^{2–6}

The first cases were reported by Frank et al⁷ and ter Haar et al.⁸ These cases were considered a variation of Melnick–Needles syndrome but were classified as a different entity called Frank–ter Haar syndrome because of the accompanying congenital cardiac defects and autosomal recessive inheritance pattern.²

Therapeutic actions are usually based on the symptoms: ophthalmology, neurology, cardiology, genetics, physiotherapy, orthopaedics and rehabilitation. Dentomaxillofacial assessment should also be included because of several oral complications.⁵ There are two reports on the oral and maxillofacial findings in Frank–ter Haar syndrome, by Parker et al⁶ and Haznedaroglu et al,⁹ and this report is the third.

This report presents the oral and maxillofacial findings of a patient with Frank–ter Haar syndrome.

Case report

A 21-year-old patient, diagnosed with Frank–ter Haar syndrome at 6 years of age, was referred to the Oral and Maxillofacial Radiology Department, Faculty of Dentistry, Istanbul University (Istanbul, Turkey), for dental complaints. Extraoral examination revealed short stature, subocular folds, full cheeks, hypertelorism, closed fontanelle, a prominent forehead and eyes, flexion deformities of the fingers, broad mouth and anteverted nostrils (Figure 1). The patient had a history of mitral valve defects, megalocornea and kyphoscoliosis.

Gingival hypertrophy, broad alveolar crests, anterior open bite and micrognathia were seen intraorally (Figure 2). The panoramic radiograph showed the following characteristics: bilateral condylar anomaly, a large right coronoid process, elongated and widened styloid processes, deep antegonial notches and multiple impacted teeth. Some impacted teeth appeared to be associated with dentigerous cysts. All of the second and third molars and lower first molars were impacted. The mandibular second and third molars were horizontally impacted (Figure 3).

CBCT revealed the absence of the frontal and sphenoid sinuses (Figure 4). The styloid processes were

Figure 1 The patient had full cheeks, micrognathia, hypertelorism, a prominent forehead and eyes, a broad mouth, subocular folds and anteverted nostrils.

hypertrophic, mandibular foramina appeared enlarged, and maxillary and ethmoid sinuses were hypoplastic bilaterally. Both mandibular condyles were flattened

Figure 2 Gingival hypertrophy, anterior open bite and broad alveolar crests seen in intraoral view.

Figure 3 Panoramic radiograph showing condylar flattening, impacted teeth, radiolucencies around the lower second and third molars, elongated and hypertrophic styloid processes, and deep antegonial notches.

and associated with a thickened roof of the glenoid fossas (Figures 5–7). The large coronoid process was seen at the right side (Figures 8 and 9).

The patient was surgically treated under general anaesthesia to remove the impacted teeth.

At the preanaesthetic examination, the patient was 120 cm tall and weighed 35 kg. Mitral and aortic regurgitation and Mobitz Type I atrioventricular block were present, and the patient was taking enalapril and metoprolol. A preoperative cardiology consultation suggested no specific precautions for the patient.

All impacted teeth were extracted in four different sessions under general anaesthesia. Based on the different texture of the oral epithelium at the lips, a minor salivary gland biopsy was performed. Healing was uneventful after the surgeries. The radiolucencies around the impacted teeth were diagnosed as inflamed hyperplastic dental follicles histopathologically, except the left mandibular area and left maxillary second molar area. The radiolucencies around these teeth were diagnosed as dentigerous cysts (Figure 10). Squamous metaplasia was seen in some regions of the odontogenic epithelium of the follicle

Figure 4 Absence of the frontal and sphenoid sinuses in the sagittal view.

Figure 5 Enlarged mandibular foramina seen in an axial slice.

walls. The minor salivary gland biopsy revealed chronic sialoadenitis histopathologically.

Discussion

Frank-ter Haar syndrome is a rare genetic disorder that results in brachycephaly, a large anterior fontanelle, a prominent forehead, hypertelorism, prominent eyes, megalocornea with or without glaucoma, full cheeks and flexion deformities at the fingers.¹⁰ All of the above mentioned features of the syndrome were also observed in our case, except for the open anterior fontanelle. We believe our patient's fontanelle was closed due to his age.

The findings in our case report are similar to the ones described earlier on Frank-ter Haar syndrome: broad mouth, broad alveolar ridges, micrognathia, delayed dental development, delayed eruption, impacted teeth, taurodont teeth, an increased gonial angle, an accentuated gonial notch, reduced condyle development, flattened condylar head, Class III malocclusion, anterior open bite, hypoplasia of the teeth,

Figure 6 Hypoplasia of the maxillary and ethmoid sinuses.

Figure 7 Flattened condylar heads and agenesis of the sphenoid sinuses.

full cheeks, gingival hyperplasia and deep palate.^{2,5,6,10} Additionally in our case, thickened roof of glenoid fossas, a large right coronoid process and follicular hyperplasias were seen.

In patients with Frank-ter Haar syndrome, the mandibular second molars tend to be impacted horizontally, as found in the two previous reports and in our patient.^{6,9} Parker *et al*⁶ and Haznedaroglu *et al*⁹ did not mention that the follicular width at the impacted second mandibular molars tended to be increased, although this was the case in both of their patients and in our patient.

Sinus underdevelopment or agenesis affects mainly the frontal sinuses; sphenoid sinus involvement is extremely rare.¹¹ Besides that, it is mentioned in the literature that, sinus agenesis or hypoplasia is seen in syndromes of craniosynostosis, osteodysplasia (Melnick-Needles syndrome) and Down syndrome.¹² Our patient had agenesis of both the sphenoid and frontal sinuses and underdeveloped ethmoid and

Figure 8 Hypertrophic and elongated right styloid process (a) and left styloid process (b).

Figure 9 Thickened roof of glenoid fossa bilaterally (a,b) and large right coronoid process (a).

maxillary sinuses. There is no information about the paranasal sinuses of patients with Frank-ter Haar syndrome in the literature.

The minor salivary gland biopsy showed chronic sialoadenitis, which has not been reported earlier in Frank-ter Haar syndrome.

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Figure 10 Hyperplastic dental follicle. Follicular epithelium that lay on the fibrotic wall is seen [haematoxylin and eosin (H&E) ×200] (a). Dentigerous cyst. Non-keratinized multilayered epithelium that lay on the fibrotic cyst wall is seen (H&E ×400) (b).

In conclusion, Frank-ter Haar syndrome is a rare syndrome that involves various maxillofacial anomalies. This report outlines some of the anomalies seen in a patient with Frank-ter Haar syndrome, but additional data about these patients' maxillofacial anomalies and dental approaches are needed.