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CASE REPORT

Common dental features and craniofacial development of patients with Ter Haar syndrome

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Abstract
Background Ter Haar syndrome is a rare genetic syndrome with <30 cases reported worldwide. There is nothing within the published literature regarding the dental development and dental features of these patients.
Case report This case series examines three patients with Ter Haar syndrome and tracks their dental development and identifies common dental and skeletal features.
Follow up All three patients received dental treatment and regular follow-up at Great Ormond Street Hospital Dental Department.
Conclusion These patients have many common dental and craniofacial features which poses the question as to whether these features are due to Ter Haar syndrome.

Keywords Ter Haar syndrome · Dental development · Craniofacial

Background

Frank-Ter Haar syndrome was first recognised by Frank et al. (1973) and later confirmed by Ter Haar et al. (1982). It is a rare genetic syndrome characterised by multiple skeletal abnormalities, developmental delay, cardiac defects and characteristic facial features (Saeed et al. 2011). The main clinical features are brachycephaly, wide fontanelles, prominent forehead, hypertelorism, prominent eyes, macrocornea with or without glaucoma, full cheeks, small chin, bowing of the long bones and flexion deformities of the fingers (Maas 2004).

It is transmitted as an autosomal recessive disorder (Femitha et al. 2012) with the most common defect being a mutation in the *SH3PXD2B* gene on chromosome 5q35.1 (Bendon et al. 2012). Less than 30 cases have been reported worldwide.

The dental features of patients with Ter Haar syndrome have not previously been reported. Little is known about the dental development of these patients. The following case series describes the dental features and dental development of three siblings with Ter Haar syndrome.

Case report

Three patients with Ter Haar syndrome have been treated at Great Ormond Street Hospital since infancy. Diagnosis of Ter Haar syndrome was made through genetic screening revealing a small deletion of the final exon on the gene *SH3PXD2B*. They are currently the only patients with Ter Haar syndrome at the hospital and attend Great Ormond Street Hospital Dental Department for their dental treatment.

Patient one

A 15-year-old male.

Medical history

Ter Haar syndrome with associated mitral and aortic valve regurgitation and cardiomyopathy. A mitral valve repair

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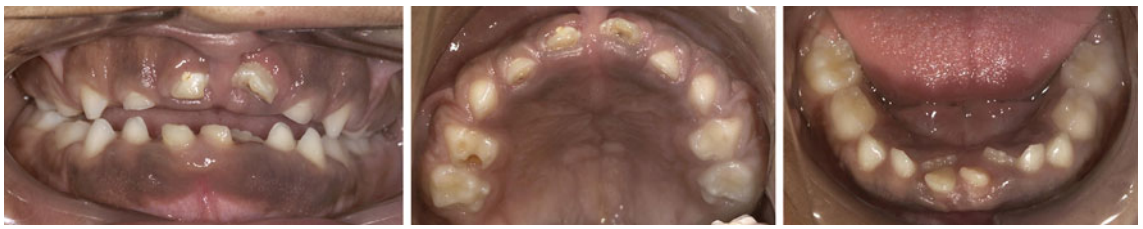

Fig. 1 Clinical photographs aged 8 years

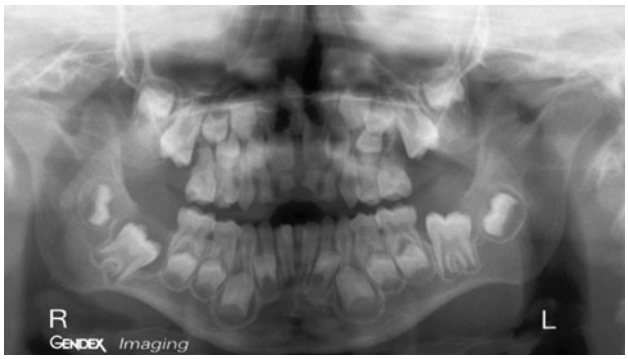

Fig. 2 DPT radiograph aged 8 years

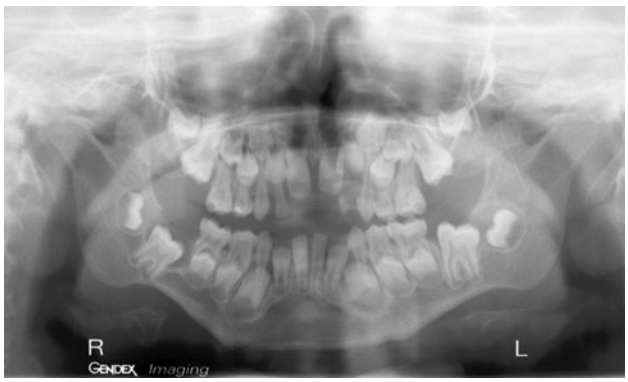

Fig. 3 DPT radiograph aged 9 years

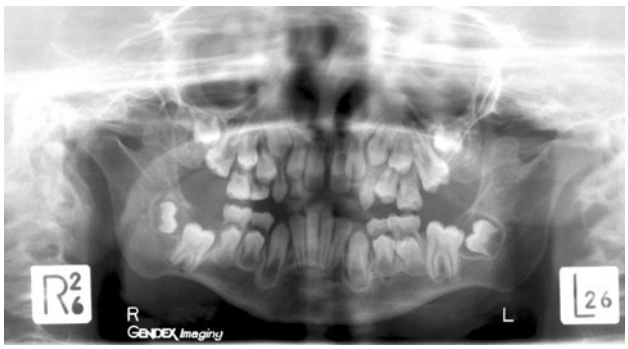

Fig. 4 DPT radiograph aged 10 years

was performed in 2010. The patient has residual left ventricular dysfunction and hearing loss in the left ear for which he wears a hearing aid.

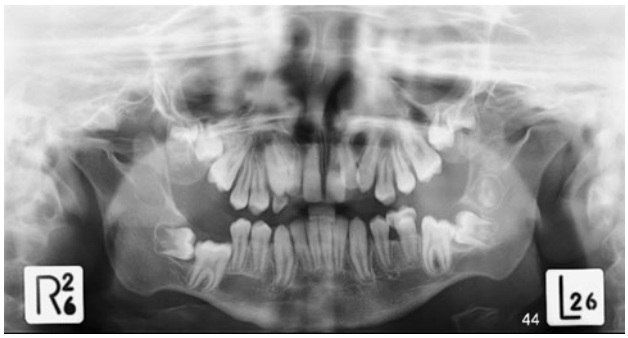

Fig. 5 DPT radiograph aged 13 years

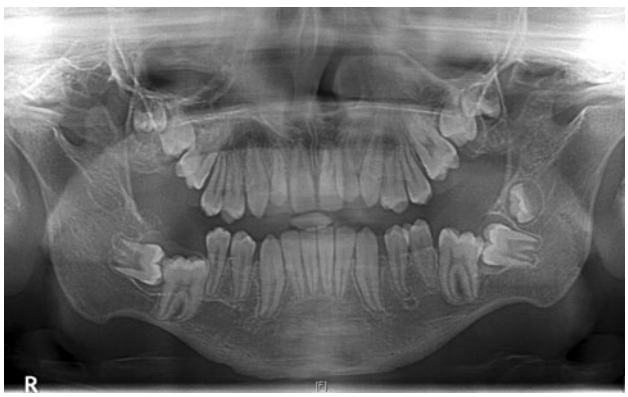

Fig. 6 DPT radiograph aged 14 years

Dental features and dental development

The main dental features exhibited by this patient are delayed dental development, generalised gingival hyperplasia and multiple impacted teeth. These are illustrated in the clinical photographs and the radiographs shown in Figs. 1–7.

Aged 8 years

In the upper and lower arch, all primary teeth were present and the lower permanent central incisors were partially erupted with the primary predecessors remaining in situ. Generalised gingival hyperplasia can clearly be seen in all photographs with minimal clinical crown height present above the gingival margins (Fig. 1).

A dental pantomograph (DPT) (Fig. 2) clearly shows delayed dental development with the permanent upper and lower incisors and upper and lower first and second molars still forming and not being close to eruption.

Aged 9 years

Permanent incisors and first permanent molars remain unerupted. The DPT in Fig. 3 shows that the patients dental development was progressing, albeit slowly.

Aged 10 years

At age 10 years, the upper permanent central incisors remained unerupted but were palpable labially and the lower permanent central and lateral incisors were partially erupted. Root formation of the first permanent molars was close to completion. The teeth were not close to eruption, especially the lower right first molar which was close to the lower border of the mandible (Fig. 4).

Aged 13 years

Clinical examination at the age of 13 years revealed the upper permanent central incisors to be partially erupted, all four lower permanent incisors to be fully erupted and all the premolars to be erupted with the exception of the lower left second premolar. The upper right primary canine was still present at this time. The upper left primary canine had been exfoliated, but the upper left permanent canine remained unerupted.

Figure 5 shows the continued dental development of the patient and the lower left second permanent molar being close to eruption. Both upper permanent canines appear to be close to eruption. The radiograph shows that the wisdom teeth are developing with the exception of the lower right third molar. Lower first permanent molars remain unerupted with the apices of the lower right tooth being close to the lower border of the mandible.

The lower right second permanent molar was seen to be developing horizontally and the lower left counterpart developing mesially, both of which were impacting on the distal aspect of the corresponding first permanent molars. The upper first and second permanent molars also appeared to be impacted, with the second molars similarly impacting. From this radiograph, it can also been seen that both upper first permanent molars exhibit taurodontism.

Clinical examination later in the year revealed the upper permanent central incisors to have erupted and the upper permanent canines and the upper permanent first molars to be buccally palpable. The upper permanent lateral incisors were unerupted and impalpable.

Aged 14 years

The permanent molars, upper canines and upper laterals were still unerupted. A DPT taken at this age can be seen in Fig. 6.

This shows the lower first permanent molars to be almost fully developed, but unerupted. The lower right first permanent molar was deeply vertically impacted with the root apices close to the lower border of the mandible. The lower right second permanent molar was horizontally impacted against the distal aspect of the first permanent molar. The lower left first permanent molar was also vertically impacted, although not as deeply as on the right. The lower left second permanent molar was mesially impacted against the distal aspect of the first permanent molar.

The upper first and second molars on both sides were seen to be developmentally delayed, with the second permanent molars impacted against the distal aspect of the first permanent molars.

Aged 15 years

A few months later, the lower right second premolar to the lower left second premolar were present with the exception of the upper left lateral incisor. Clinical photographs (Fig. 7) showed the patient's delayed dental development and gingival hyperplasia which were especially evident in the upper anterior region, where the upper left three had only minimal height crown present above gingival level and the upper left two was imbedded within hyperplastic gingiva.

Skeletal features

In addition to the dental features discussed, the series of DPT radiographs showed the patient to have an increased gonial angle, an accentuated gonial notch and reduced

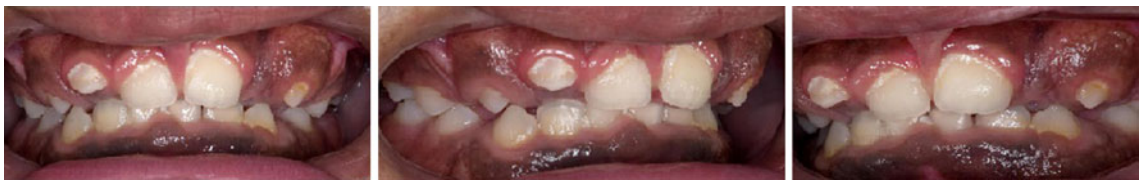

Fig. 7 Clinical photographs aged 15 years

development and flattening of the condylar heads. Clinical examination revealed a skeletal three pattern of growth.

Patient two

A 10-year-old female.

Medical history

Ter Haar syndrome with associated mild mitral valve regurgitation, trace regurgitation of the aortic valve and unequal foot size. In April 2010, she experienced increased intra-cranial pressure due to craniosynostosis for which calvarial expansion was successfully performed.

Dental features and dental development

The main dental features are the same as for patient one: delayed dental development, generalised gingival hyperplasia and multiple impacted teeth. Her dental development also followed the same pattern. The dental features can be seen from the clinical photographs and radiographs shown in Figs. 8, 9.

Aged 3 years

All upper and lower primary teeth, up to the first primary molars, were present. There was generalised gingival hyperplasia especially in the lower anterior region, with little clinical crown height visible above gingival level (Fig. 8). A polyp was present on the lower right labial mucosa of 4 mm diameter.

Aged 9 years

The permanent upper central incisors remained unerupted and embedded within hyperplastic gingivae which also affected the patient's speech. All first permanent molars were also unerupted at this time. All four first premolars were partially erupted. This was much earlier than the other two patients described in this report.

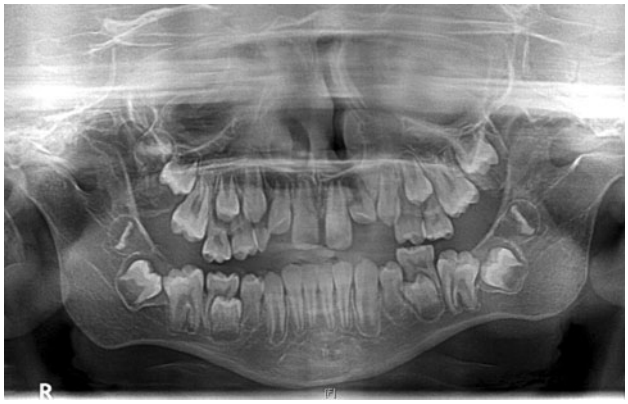

Fig. 9 DPT radiograph aged 9 years

A DPT (Fig. 9) shows the patients developing dentition and illustrates the delayed development, especially of the permanent molars. The roots of the first permanent molars were still developing and the teeth were not close to eruption, especially in the mandible. The lower second molars were seen to be mesially inclined and appeared to be beginning to impact on the distal aspect of the lower first molars. The upper first permanent molars were seen to be taurodonts.

The features seen clinically and radiographically at this age correspond to the features seen in patient one.

Skeletal features

An increased gonial angle, accentuated gonial notch and reduced development and flattening of the condylar heads. These findings correspond with those of patient one. Clinically, this patient also has a skeletal three pattern of growth.

Patient three

A 9-year-old male.

Medical history

Ter Haar syndrome with no additional medical conditions.

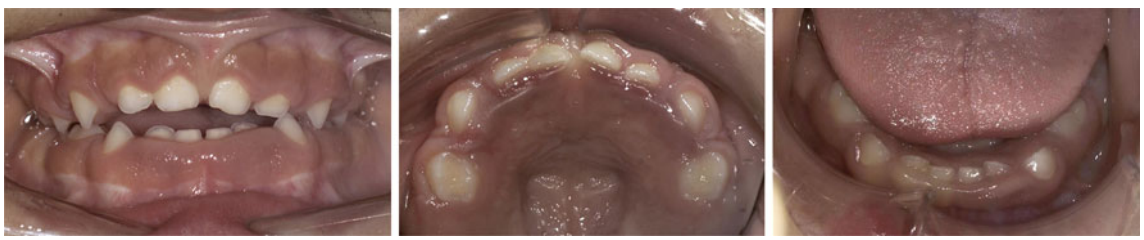

Fig. 8 Clinical photographs aged 3 years

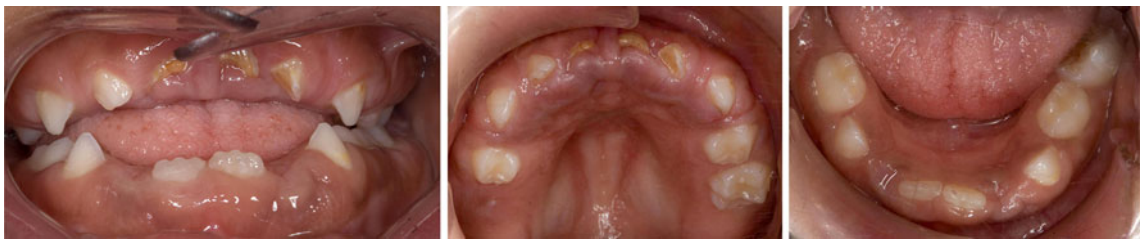

Fig. 10 Clinical photographs aged 7 years

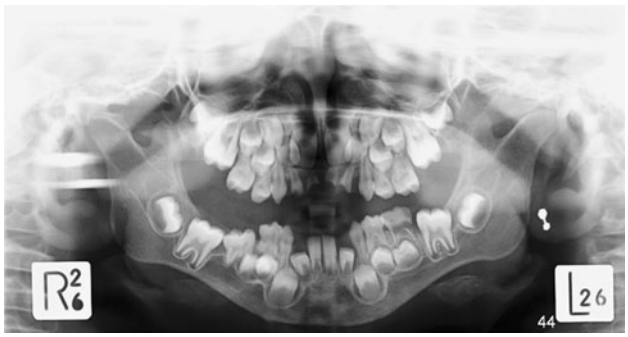

Fig. 11 DPT radiograph aged 7 years

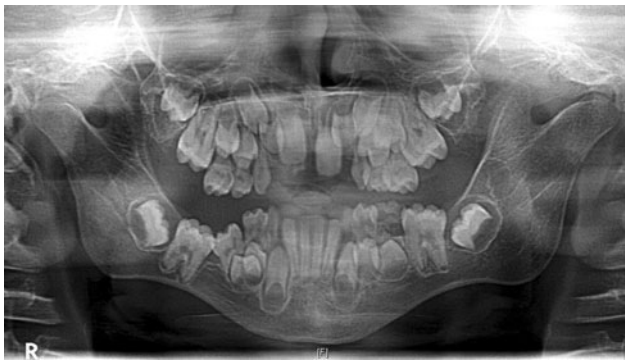

Fig. 12 DPT radiograph aged 8 years

Dental features and dental development

The dental features of this patient are consistent with those of the other two patients, showing delayed dental development, gingival hyperplasia and impacted teeth. These features can be seen in clinical photographs and radiographs in Figs. 10–12.

Aged 7 years

In the maxillary arch, the upper right first primary molar to the upper left second primary molar were erupted. In the mandibular arch, the lower right primary first molar and canine and lower left primary second and first molars and canine were present, with the permanent lower incisors partially erupted. The eruption of the lower central incisors was seen earlier in this patient than the other two patients. The patient also had generalised gingival hyperplasia, similar in nature to the other two patients.

Clinical photographs (Fig. 10) show that the upper central and upper left primary lateral incisor have caries. Gingival hyperplasia can clearly be seen with the gingiva growing over parts of the upper anterior teeth. All teeth having minimal clinical crown height shown above the gingival margins.

In addition to the clinical findings, DPT taken at this age (Fig. 11) shows that the lower left primary second molar has internal resorption and that the lower right primary second molar was present but submerged close to the level of the crestal bone. The first permanent molars were present but were much delayed in their development and were not close to erupting. Lower second permanent molars were developing with a mesial inclination. These findings were similar to those seen in the other two patients, especially with regards to delayed dental development of the first permanent molars and the mesial inclination and impaction of the lower second permanent molars.

Aged 8 years

Clinical examination revealed that the permanent upper central and lateral incisors were still unerupted and no permanent molars had erupted.

A DPT (Fig. 12) illustrates the delayed dental development with the first permanent molars not yet fully developed. The lower right first permanent molar was infra-occluded. The lower second molars were continuing to develop, whilst remaining mesially inclined with the appearance of beginning to impact on the distal aspect of the lower first permanent molars. The upper first permanent molars were appearing to impact and the second permanent molars showed delayed development. The upper first permanent molars were also seen to be taurodonts.

Aged 9 years

Lower left first permanent molar was partially erupted. All other first permanent molars and upper central incisors remained unerupted.

Table 1 Common dental and skeletal features

Feature	Patient 1	Patient 2	Patient 3
Gingival hyperplasia	+	+	+
Delayed dental development	+	+	+
Delayed eruption	+	+	+
Impacted teeth	+	+	+
Taurodont teeth	+	+	+
Mucosal polyp	–	+	–
Increased gonial angle	+	+	+
Accentuated gonial notch	+	+	+
Reduced condyle development	+	+	+
Flattened condylar head	+	+	+
Skeletal base class	3	3	3

Skeletal features

Radiographs demonstrated an increased gonial angle, accentuated gonial notch and flattening of the condylar heads. Clinical examination confirmed a skeletal three pattern of growth.

Discussion

The three cases described in this series showed similar dental and skeletal features and similar dental development. All had generalised gingival hyperplasia, delayed dental development, especially of the permanent molars and delayed dental eruption along with the impaction of multiple teeth. In addition to late development of the incisors, their eruption appeared to be delayed by the presence of the hyperplastic gingiva. Another common finding was the pattern of development of the lower second permanent molars where their mesial path of eruption caused them to impact against distal aspect of the lower permanent first molars.

Skeletally all three patients had an increased gonial angle, an accentuated gonial notch, flattening of the condylar heads along with a skeletal three growth pattern.

These common findings are summarised in Table 1.

Although these patients are siblings, neither their non-Ter Haar parents nor their unaffected siblings had similar dental problems. We therefore propose that these findings are not due to simple familial similarity, but rather were due to their Ter Haar syndrome. There are no published studies on the dental and skeletal features of patients with Ter Haar syndrome; therefore, it is not possible to compare these findings with the findings of other, unrelated Ter Haar syndrome patients.

Conclusion

Ter Haar syndrome is an interesting and rare syndrome with little published literature. No literature exists on the dental features and dental development of patients with this syndrome. Due to rarity of the condition, it will be difficult to analyses these features in a large number of patients. This case series is the first literature of its type and attempts to fill this gap.

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