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Is amelogenesis imperfecta an indication for renal examination?

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Background. The term 'amelogenesis imperfecta' (AI) describes a diverse group of hereditary conditions primarily affecting the quality and/or quantity of dental enamel.

Case report. This paper describes a case in which hypoplastic AI with delayed/failure of eruption of the permanent teeth was shown to be associated with renal calcification.

Conclusion. Given the importance of the renal involvement, the authors suggest that paediatric dentists consider referring all children with this dental phenotype for renal ultrasound examination.

Introduction

The term 'amelogenesis imperfecta' (AI) describes a diverse group of hereditary conditions primarily affecting the quality and/or quantity of dental enamel. Amelogenesis imperfecta may affect all or only some of the teeth in the primary and/or permanent dentition. Its mode of inheritance may be X-linked, autosomal dominant or autosomal recessive, although, where the affected individual is the first family member to present, this can be difficult to elicit; prevalence varies with the population group studied[1–4], having been quoted as between 1:14 000 and 1.4:1000.

Although AI is generally considered to primarily affect dental enamel, other oral and dental stigmata (including unerupted teeth, anterior open bite, pulpal calcifications, interradicular dentinal dysplasia, root and crown resorption, cementum deposition, truncated roots, and taurodontism[5–14]) have been shown to coexist. In addition, AI is a feature of several multiorgan syndromes, but is pathognomonic of only a few.

In the past 30 years, an extremely rare syndrome associating AI with nephrocalcinosis (OMIM 204690) has been reported in just a few families[15–20]. In reporting a further case, the authors' aim is to raise paediatric dentists' awareness of this potential association in order that affected individuals might benefit from early referral to nephrology services and hence improved prognosis.

Case report

A Caucasian male aged 13.9 years, was referred to a multidisciplinary clinic at the University Dental Hospital, Cardiff, UK, regarding failure of eruption of the majority of his permanent dentition. He had a confirmed diagnosis of hypoplastic AI and had previously been treated at a local district general hospital's restorative dentistry unit; a panoramic radiograph taken at the age of 9.8 years was available to the multidisciplinary team. The child's previous management had included the modification of a number of primary teeth with composite resin and the provision of a maxillary overdenture. An only-child, he had no relevant medical history, and there was no family history of either amelogenesis imperfecta or failure of eruption of permanent teeth.

On presentation, intraoral examination revealed the maxillary left central incisor and the mandibular central incisors to be the only erupted permanent teeth; the mandibular central incisors were divergent, occupying the normal position of the permanent mandibular lateral incisors. The retained primary and erupted permanent teeth all showed a yellow

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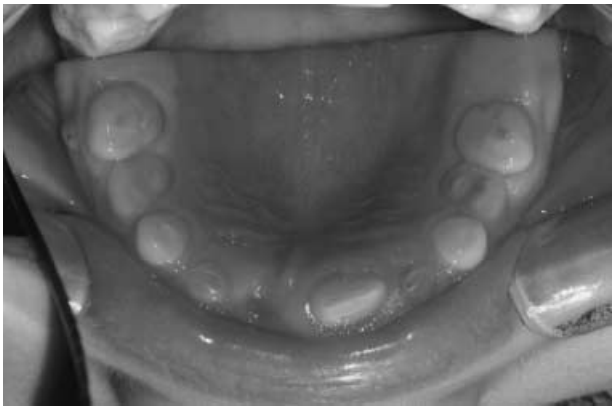

Fig. 1. Occlusal view of the maxillary arch.

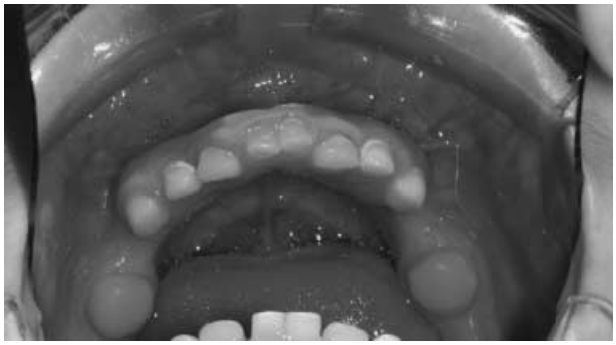

Fig. 2. Occlusal view of the mandibular arch.

discoloration, with thin enamel. In response to this latter observation, the divergent mandibular permanent central incisors had been restored with composite. In the maxillary arch, the primary molars, canines and lateral incisors were retained and exhibited advanced wear, the molars being devoid of any cuspal features. In the mandibular arch, the primary central incisors, canines and second molars were present, having been previously modified with adhesive restorations (Figs 1 & 2).

Scrutiny of the panoramic radiograph (Fig. 3) taken at age 9.8 years revealed that, at that stage, his first permanent molars had failed to erupt, and the maxillary left central incisor and both mandibular central incisors were the only permanent anterior teeth present in the mouth. Interestingly, the lower primary central incisors were retained and the first primary molars infraoccluded. The mandibular second premolars were ectopically placed, being located inferior to the mesial roots of their predecessors. Crown morphology, particularly that of the developing dentition, was noted to be abnormal, the density of the enamel appearing very similar to that of the dentine.

Comparison of the panoramic radiograph taken at presentation to the multidisciplinary clinic (Fig. 4) with the film taken approximately 4 years previously revealed that, while the roots of the teeth in the permanent series had continued to form, there had been little or no axial movement through bone in the intervening period. On the contrary, there was bone

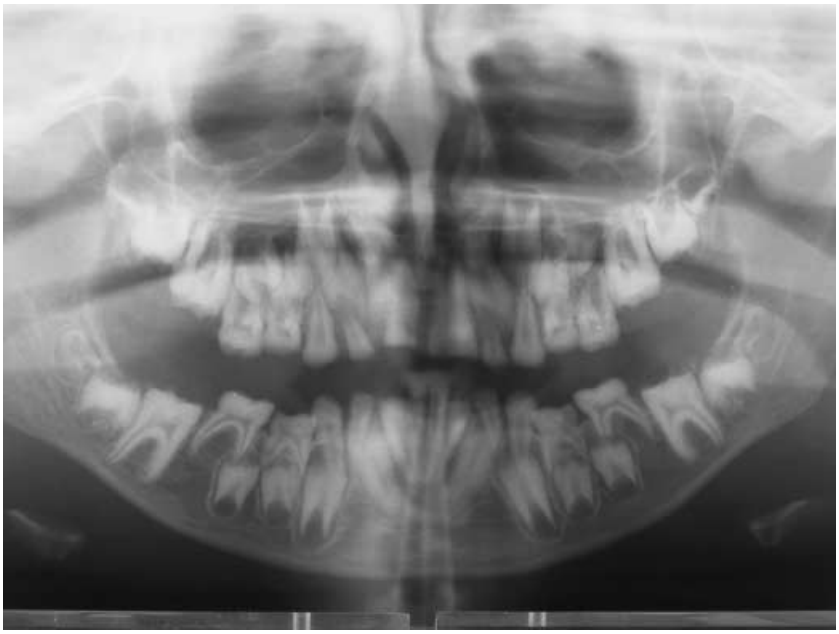

Fig. 3. Dental panoramic tomograph taken at 9.8 years of age.

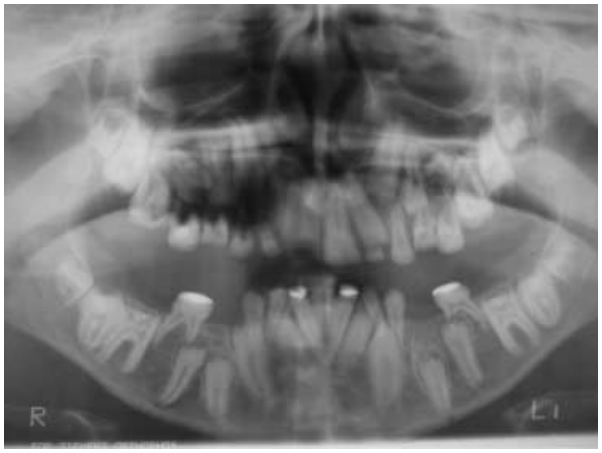

Fig. 4. Dental panoramic tomograph taken at 13.9 years of age.

of normal density covering the occlusal surfaces of the mandibular first permanent molar teeth. These teeth exhibited pulp stones and there was some evidence of internal resorption within their pulp chambers. Finally, the maxillary left second premolar appeared odontome-like. In interpreting this radiograph, it should be noted that the extensive radiolucency in the maxillary right quadrant is artefactual.

In the course of preparation of this case for publication, the authors conducted an electronic literature search on the keywords 'amelogenesis imperfecta' and 'delayed eruption'. This revealed an extremely rare association of AI with nephrocalcinosis. As a precautionary measure, therefore, he was referred to a consultant in paediatric nephrology at the KRUF Children's Kidney Centre for Wales. Ultrasound examination of his kidneys and bladder demonstrated renal calcification in a medullary distribution consistent with a diagnosis of mild nephrocalcinosis. Subsequent haematological examination revealed a marginally elevated serum creatinine (112 μmol L^{-1}); the estimated glomerular filtration rate was 65 mL min^{-1} per 1.73 m^2. The child was noted to have mild proteinuria, but no abnormalities of parathyroid hormone or urine calcium. He has been advised to lead a healthy lifestyle with reasonable levels of exercise and a reduction in calorie intake, his weight currently being above the ninety-seventh centile. It is intended that he will undergo routine renal review on a 6-monthly basis.

From a dental viewpoint, the management of patients presenting with primary failure of eruption is extremely difficult, since the teeth involved tend to ankylose when orthodontic traction is applied. In the absence of a normal response to orthodontic force, the only way to move unerupted teeth into occlusion is to reposition them surgically without disturbing the periodontal ligament. When this is not feasible, prosthetic replacement of the abnormal teeth becomes the only treatment possible. In the present case, a combined surgical and orthodontic approach was considered to bring the unerupted teeth into alignment. However, the limited vertical anchorage and suspected ankylosis of the unerupted teeth contraindicated this. While localized osteotomies and distraction osteogenesis are a theoretical possibility for the future, he will continue to receive prosthodontic maintenance until growth is complete.

Discussion

The first report of this syndrome (occurring in a sibling pair) appeared in 1972[15]. One sibling died at the age of 26 years, having suffered severe renal failure as a complication of nephrocalcinosis; the other developed multiple urinary infections, hypertension and renal failure. Subsequently reported cases[16–20] share the following common features: failure of eruption, enamel agenesis, unexplained nephrocalcinosis, and normal plasma calcium, 25-OH vitamin D3, alkaline phosphatase and parathyroid functions.

Given the small number of reported cases to date, the prognosis associated with this syndrome has not been established, though unrecognized and untreated nephrocalcinosis is known to be associated with significant morbidity. In some of the previous case reports[18,19], it has been suggested that children with apparently autosomal recessive AI should, at least, have a renal ultrasound examination to exclude such pathology. Going a stage further, Paula and co-workers[20] have advocated that all patients with AI should undergo such an examination.

Regrettably, only one of the previous publications concerning this syndrome has appeared in a journal specifically targeted at the very group of professionals who are best placed to ensure that potentially affected children benefit from early referral. As paediatric dentists, we consider that further information is urgently

required as to whether this significant renal pathology is exclusive to autosomal recessive hypoplastic AI or whether it can be found in association with other AI variants; investigation at the molecular level is to be encouraged.

What this paper adds

- This case report adds a new case of an extremely rare syndrome to the dental literature.
- With one exception, previous reports of this syndrome have not been specifically targeted at paediatric dentists.

Why this paper is important to paediatric dentists

- Unrecognized and untreated nephrocalcinosis is associated with significant morbidity. Paediatric dentists should be aware of the possible association of failure of tooth eruption and hypoplastic AI with renal calcification, and refer affected children appropriately.
- Further research is required in order to determine whether the association occurs in other AI variants.

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