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Clinical Observation

Amelogenesis imperfecta: signs that should alert pediatric dentists

Najla Taktak[1,*], Lamia Mansour[1], Sameh Sioud[2]

[1] Service de Prothèse partielle amovible, Clinique hospitalo-universitaire de Médecine dentaire, Monastir, Tunisie
[2] Service de Médecine et Chirurgie buccales, Clinique hospitalo-universitaire de Médecine dentaire, Monastir, Tunisie

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Abstract – This article describes a new case of a rare syndrome which combines uncommon conditions, such as hypoplastic amelogenenesis imperfecta (AI), delay of permanent tooth eruption, gingival enlargement, pulpal calcifications and bilateral medullary nephrocalcinosis. The importance of syndrome diagnosis and recognition in this condition is in guiding pediatric dentist, who meets this patient group in early age, to recognize the possibility of renal anomalies in patients AI in order that affected individuals might benefit from early referral to nephrology services and hence improved prognosis.

Mots clés :
amélogenèse imparfaite / néphrocalcinose

Résumé – Amélogénèse imparfaite : signes d'alerte pour les pédodontistes. L'amélogenèse imparfaite est une anomalie de la structure de l'émail qui peut toucher les deux dentures. Elle est souvent associée à un retard d'éruption et à des inclusions multiples. Ce tableau clinique bucco-dentaire peut être associé à une néphrocalcinose médullaire qui peut évoluer vers l'insuffisance rénale.
Cet article a pour objectif de sensibiliser les médecins dentistes sur l'intérêt du dépistage précoce du syndrome associant l'amélogenèse imparfaite à la néphrocalcinose afin d'améliorer le pronostic vital des patients.

Amelogenesis imperfecta (AI) is a diverse group of hereditary conditions that affects the quality and quantity of dental enamel [1]. It may affect all or only some teeth in the primary and /or permanent dentition [2]. Inheritance is mainly autosomal dominant, but autosomal recessive, X-linked and sporadic cases can also occur spontaneously in one or more members of the same family [3]. AI also occurs as an integral and often diagnostic feature of a small number of syndromes [4]. A rare syndrome associating amelogenesis imperfecta with nephrocalcinosis (OMIM 204690), precipitation of calcium salts in the renal tissue, has been reported in just a few families [5]. In reporting a further case, the authors aim is to raise pediatric dentists awareness of this potential association.

Case report

A 19-year-old female patient was referred to the department of prosthetic dentistry for esthetic reasons than for functional reasons. The patient's parents were first cousins in first degree. Her father and brother had dental anomalies. Examination revealed no relevant medical history, apart from her dentition, and general development was normal.

Intraoral examination revealed the retained primary and erupted permanent teeth all showed alterations in the tooth shape with yellow discoloration, thin enamel and large interproximal spaces (Fig. 1). In the maxillary arch, the primary canines were retained. In the mandibular arch, the primary canines, right first molar and left second molar were present (Fig. 2). She had a slight gingival enlargement but had no anterior open bite. Panoramic radiograph revealed the presence of second and third molars which were clinically absent in all four quadrants. No density difference between enamel and dentin was observed. The unerupted permanent canines and mandibular second premolars were ectopically placed and had large well-defined pericoronal radiolucencies. Finally, it should be noted agenesis of the mandibular left canine and coronal intrapulpal calcifications in all permanent first molars (Fig. 3). The clinical and radiographic features led to the diagnosis of AI, hypoplastic type and appropriate cosmetic rehabilitation was carried out. Investigations revealed nephrocalcinosis on X-ray film of the abdomen and

* Correspondence: farfattoutn@yahoo.fr

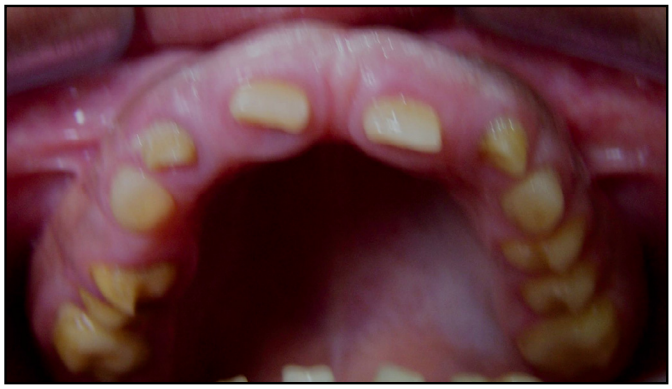

Fig. 1. Maxillary arch.
Fig. 1. Arcade dentaire maxillaire.

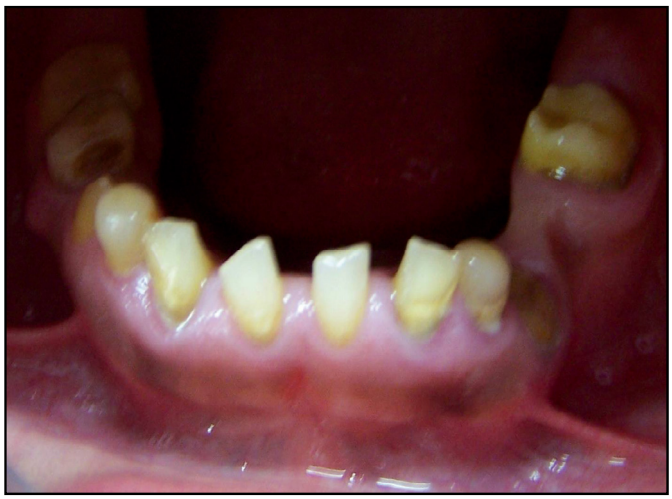

Fig. 2. Mandibular arch.
Fig. 2. Arcade dentaire mandibulaire.

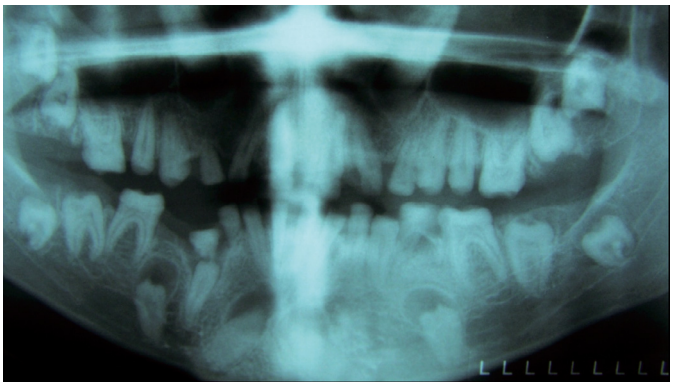

Fig. 3. Panoramic radiograph.
Fig. 3. Radiographie panoramique.

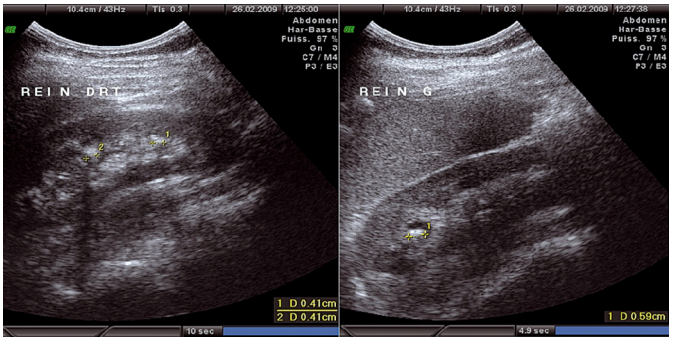

Fig. 4. Ultrasound showing medullary kidney calcifications.
Fig. 4. Echographie montrant des calcifications rénales.

was confirmed by ultrasonography which showed renal calcification in a medullary distribution consistent with a diagnosis of bilateral medullary nephrocalcinosis (Fig. 4). Subsequent haematological examination revealed no disturbance in calcium metabolism or excretion, and renal function was normal. Blood electrolytes, serum urea, creatininemia and proteinuria levels were all normal.

Discussion

AI has been classified by Witkop four major types of AI based on phenotype, namely hypoplastic, hypocalcified, hypomaturation and hypomaturation-hypoplastic types are currently recognized [1]. This classification based primarily on phenotype was considered unsatisfactory. The recent workable classification proposes that the mode of inheritance be considered as the primary factor in the diagnosis of AI, followed by the gene mutation, the biochemical outcome if known and finally the phenotype [6]. To date, mutations in four genes (AMELX, ENAM, KLK4 and MMP 20) have been reported to cause AI [7]. In the present case, the consanguineous of the patient's parents suggests an autosomal recessive inheritance.

In some of the previous case reports [4,8–13], it has been suggested that children with apparently autosomal recessive AI should, at least, have a renal ultrasound examination to exclude the combination of AI and nephrocalcinosis. The AI and nephrocalcinosis syndrome has been reported in consanguineous and non consanguineous families [12]. In 1972, Mac Gibbon reported a first sibling pair with autosomal recessive hypoplastic AI and nephrocalcinosis in a non consanguineous family. The brother died at the age of 26, with a severe renal failure as a complication arising from his nephrocalcinosis. The sister also developed multiple urinary infections, hypertension and renal failure [8]. This syndrome of AI and nephrocalcinosis is characterized by delayed tooth eruption, the presence of thin or absent enamel, presence of intrapulpal calcifications and bilateral medullary nephrocalcinosis with normal calcemia [8–13]. The delay of eruption could be explained by the pathology [14, 15] or the presence of some calcifications in the dental follicles [11]. The presence of abnormal enamel and intrapulpal calcifications suggest that

the tooth morphogenesis and dentinogenesis are also affected in the syndrome [12]. The syndrome of AI and nephrocalcinosis was studied by Phakey and al. [16] and Hall and al. [4]. The study suggested the possibility of an abnormality in interstitial matrix, which could lead to dystrophic calcifications in the kidney and abnormal tooth enamel formation [9]. It also suggested the possibility of involvement of two separate but closely linked genes [4]. Another hypothesis suggests that many of the dental proteins that were believed to be tissue specific may be expressed in more than one dental tissue and also in non-dental tissues, and these proteins may have a role in calcium and phosphate metabolism [3, 16–20].

Dental development disorder requires a team approach with the pediatric dentist as coordinator, an oral surgeon, a periodontist, an orthodontist, and finally a prosthodontist. This syndrome is extremely rare and the prognosis is unknown. Given the importance of the renal involvement, all patients with AI should be referred for medical examination including renal functions studies and ultrasonography to detect nephrocalcinosis and hence improved renal prognosis.

Competing interests: none

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