

Clinical Observation

Amelogenesis imperfecta: signs that should alert pediatric dentists

Najla Taktak^{1,*}, Lamia Mansour¹, Sameh Sioud²

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 amelogenesis 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 :

amélogénèse imparfaite /
néphrocalcinose

Résumé – Amélogénèse imparfaite : signes d'alerte pour les pédodontistes. L'amélogénèse 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'amélogénèse 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.

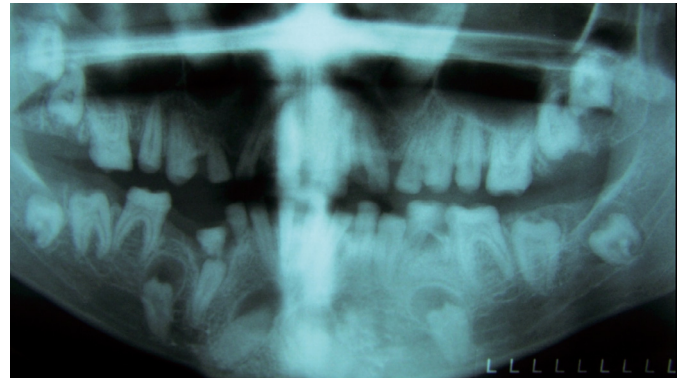


Fig. 3. Panoramic radiograph.
Fig. 3. Radiographie panoramique.

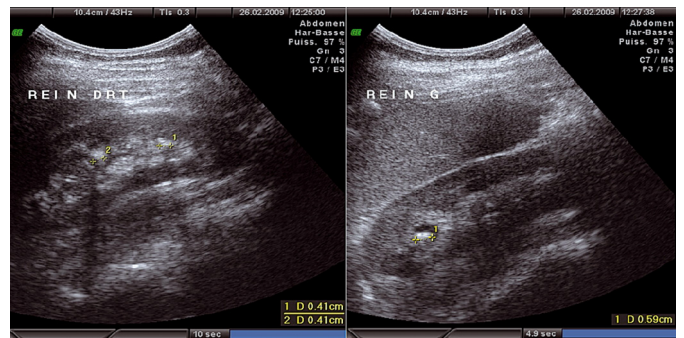


Fig. 4. Ultrasound showing medullary kidney calcifications.
Fig. 4. Echographie montrant des calcifications rénales.

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 Phahey 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

References

1. Witkop Jr CJ. Amelogenesis imperfecta, dentinogenesis imperfecta and dentin dysplasia revisited: problems in classification. *J Oral Pathol* 1989;17:547–53.
2. Hunter L, Liam D, Knox J, Drage N. Is amelogenesis imperfecta an indication for renal examination? *Intl J Paediatr Dent* 2007;17:62–5.
3. Elizabeth J, Lakshmi Priya E, Umadevi KMR, Ranganathan K. Amelogenesis imperfecta with renal disease - a report of two cases. *J Oral Pathol* 2007;36:625–8.
4. Hall RK, Phahey P, Palamara J, Mc Credie DA. Amelogenesis imperfecta and nephrocalcinosis syndrome, case studies of clinical features and ultrastructure of tooth enamel in two siblings. *Oral Surg Oral Med Oral Pathol* 1995;79:583–92.
5. OMIM (Online Mendelian Inheritance in Man) National library of medicine, Center for medical genetics, Johns Hopkins University and National Center for Biotechnology Information, Baltimore, 2004.
6. Aldred MJ, Savarirayan R, Crawford PJ. Amelogenesis imperfecta: a classification and catalogue for the 21st century. *Oral Dis* 2003;9:19–23.
7. Crawford PJ, Aldred M, Bloch-Zupan A. Amelogenesis imperfecta. *Orphanet J Rare Dis* 2007;2:17.
8. Mac Gibbon D. Generalized enamel hypoplasia and renal dysfunction. *Aust Dent J* 1972;17:61–3.
9. Lubinsky M, Angle C, Marsh P W, Witkop Jr CJ. Syndrome of amelogenesis imperfecta, nephrocalcinosis, impaired renal concentration, and possible abnormality of calcium metabolism. *Am J Med Genet* 1985; 20:233–43.
10. Dellow EL, Harley KE, Unwin RJ, Wrong O, Winter GB, Parkins BJ. Amelogenesis imperfecta, nephrocalcinosis, and hypocalcemia syndrome in two siblings from a large family with consanguineous parents. *Nephrol Dial Transplant* 1998;13:3193–6.
11. Normand TI, Bonarek H, Marteau JM, Boileau MJ, Nancy J. Amelogenesis imperfecta, nephrocalcinosis: a new case of this rare syndrome. *J Clin Pediatr Dent* 2003;27:171–5.
12. Paula LM, Melo NS, Silva Guerra EN, Mestrinho DH, Acevedo AC. Case report of a rare syndrome associating amelogenesis imperfecta and nephrocalcinosis in a consanguineous family. *Arch Oral Biol* 2005;50:237–42.
13. Kirzioglu Z, Ulu KG, Sezer MT, Yüksel S. The relationship of amelogenesis imperfecta and nephrocalcinosis syndrome. *Med Oral Patol Oral Cir Bucal* 2009;14:579–82.
14. Marks SC. The basic and applied biology of tooth eruption. *Connect Tissue Res* 1995;32:149–57.
15. Wise GE. Cell and molecular bio of tooth eruption (pp. 1–8). In *The biological mechanisms of tooth eruption, resorption and replacement by implants*. Z. Davidovitch ed: EBSCO Media, Birmingham, 1998.
16. Phahey P, Palamara J, Hall RK, Mc Credie DA. Ultrastructural study of tooth enamel with amelogenesis imperfecta in AI-nephrocalcinosis syndrome. *Connect Tissue Res* 1995 32:253–9.
17. Dellow EL, Harley KE, Unwin RJ, Wrong O, Winter GB, Parkins BJ. Amelogenesis imperfecta, nephrocalcinosis and hypocalcemia syndrome in two siblings from a large family with consanguineous parents. *Nephrol Dial Transplant* 1998;13:3193–6.
18. Deutsh D, Leiser Y, Shay B, Fermon E, Taylor A, Rosenfeld E, Dafni L, Charuvi K, Cohen Y, Haze A, Fuks A, Mao Z. The human tuftelin gene and the expression of tuftelin in mineralizing and non mineralizing tissues. *Connect Tissue Res* 2002;43:425–34.
19. Butler WT, Brunn JC, Qin C. Dentin extracellular matrix (ECM) proteins: comparison to bone ECM and contribution to dynamics of dentinogenesis. *Connect Tissue Res* 2003;44:171–8.
20. Qin C, Runn JC, Cadena E, Ridall A, Butler WT. Dentin sialoprotein in bone and dentin sialoprotein in bone and dentin sialophosphoprotein gene expressed by osteoblasts. *Connect Tissue Res* 2003;44:179–83.