Case report: clinical management of hypoplastic amelogenesis imperfecta

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Introduction

Amelogenesis imperfecta (AI) is a group of hereditary defects in the development of tooth enamel, the transmission of which is mainly autosomal dominant that characterise specific clinical variations of this genetic disorder. The autosomal dominant transmission mode, with heterozygotic carriers, has 50% transmission to progeny and a complete and typical clinical picture. There also exists the much more frequent transmission mode linked to a recessive autosomal gene and another linked to sex, X-linked [Witkop, 1973; Burzynski et al., 1973; Escobar et al., 1981; Haug and Ferguson, 1981; Bertelli et al., 1987; Backman and Holmgren, 1988].

Differential diagnosis among different types of AI. Clinical and histological characteristics differentiate the three types of AI [Uzamis et al., 1997; Seymen and Kiziltan, 2002]. On the other hand, the radiographic aspect of the dental elements exhibits a perfectly normal tooth pulp profile and radicular morphology in all clinical forms.

In AI type I (hypoplastic) the enamel exhibits density and calcification characteristics within the norm. However, the type I is associated with a more or less marked reduction of the homonymous dental tissue. Clinically, it is possible to distinguish, in relation to the phenotype and to the type of hereditary transmission, as many as 7 different forms of AI type I (Table 1) [Schroeder, 1997].

In AI type II (hypomature) [Wright and Butler, 1989] the clinical characteristics of the genetic defect may be attributed to an alteration of the physiological processes of maturation of the enamel prisms. In fact, the organic matrix of the enamel in AI type II is secreted in normal quantities so that the thickness of the enamel, in contrast to type I, is regular. However, mineralization is reduced, in that, following the initial processes of calcification, the pre-eruptive maturation process, determined by the re-absorption of the organic matrix by the ameloblasts, is not completed or does not take place at all. This defect in the structural disposition of the hydroxyapatite crystals and the rearrangement of the enamel organic matrix will mirror the clinical characteristics of this type of AI, where the enamel is of normal thickness but crumbly, liable to abrasion and flakes very easily. Also, in the hypomaturation form, it is possible to distinguish clinical varieties (Table 1) [Wright and Butler, 1989].

In AI type III (hypocalcified) an organic matrix of
the enamel is secreted of normal thickness, but it is structurally anomalous and, therefore, not permitting the regular process of mineralization. Clinically, the enamel is opaque and of a whitish-honey colour which tends to become dark brown over time [van Waes and Stockli, 2001]. The alteration of the enamel matrix hinders a normal mineralization, giving an eroded clinical appearance of the enamel. At the end of the process of maturation it will contain more than 10% of organic substances against a normal value of 1%.

Congleton and Burke [1979] have reported three cases of AI associated with taurodontism. Other authors have also noted the presence of AI and taurodontism in the tricho-dental-osseous syndrome, characterised by increased bone density, especially of the cranium, and by curly hair. Price [1998] has hypothesised that this dominant autosomal condition may be provoked by a mutation in the distal gene DLX3 of the homeo-box. Although the characters of taurodontism and AI in this condition exhibit complete penetration among those affected, these characteristics of bone and hair are expressed variably, even when the same deletion is present in a family. This indicates that other genes and/or environmental factors influence variability of expression. Within hereditary pathologies are included the various pathologic displasias of enamel such as betodermic dysplasia (a hereditary disorder due to an alteration of the embryonal ectoderm). These displasias exhibit, in certain of its forms, dysplasia of the enamel deriving from a disturbance of the ameloblastic series taking place during its period of formation [Levin, 1988; Polimeni et al., 1996]. This structural anomaly of tooth enamel occurs in the general population with a frequency ranging from 1 to 14/16,000 live births [Burzynski et al., 1973; Aldred et al., 2003; Seow, 1993] and shows a wide range of clinical aspects. The defect affects the enamel and both primary and permanent dentitions. The purpose of the present work is to emphasise, in a clinical case, the importance of a correct diagnostic and therapeutic approach to children affected by AI.

**Case report**

A nine year old female was referred for specialist paediatric dental care. From the family medical history it appeared that the patient’s father wore a fixed prosthesis because he was affected by an hereditary enamel defect. A physical intraoral examination of the child revealed an advanced mixed dentition with the presence, in the maxillary and mandibular arches, of the first permanent molars and of the lower erupting canines.

The crown morphology of teeth was slightly irregular, vestibular and occlusal surfaces of

<table>
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<tr>
<th>AI TYPE</th>
<th>CLINICAL VARIATION</th>
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<tr>
<td>Type I (hypoplastic)</td>
<td>IA pitted (AD)</td>
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<tr>
<td>IB local (AD)</td>
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<tr>
<td>IC local (AR)</td>
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<td>ID smooth (AD, highly penetrative)</td>
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<tr>
<td>IE linked to sex (XL dominant)</td>
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<tr>
<td>IF rough (AD)</td>
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<tr>
<td>IG aplastic (AR)</td>
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<tr>
<td>Type II (hypomature)</td>
<td>IIA pigmented (AR)</td>
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<td>IIB linked to sex (XL recessive)</td>
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<td>IIC snow capped teeth (XL recessive)</td>
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<tr>
<td>Type III (hypocalcified)</td>
<td>IIIA Hypocalcified (AD)</td>
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<td>IIIB Hyocalcified (AR)</td>
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*AD autosomal dominant transmission \ AR autosomal recessive transmission \ XL transmission linked to chromosome X*

**Table 1 - Clinical classification of amelogenesis imperfecta [Witkop, 1973].**
anterior and exterior sections were rough and irregular and there was extensive dental plaque (Fig. 1a). There were defects of the crowns of the incisors involving the incisal and median thirds and a greatly increased overjet (Fig. 1b). Light probing revealed hard normal enamel, but with reduced thickness. All posterior teeth were similarly affected (Figs. 2a, b). The patient’s initial orthopantomograph showed radiopacity with slight irregular crown morphology of the teeth. Based on the clinical and radiographic examination a diagnosis was made of AI with hypoplastic enamel, of rough appearance, probably with autosomal dominant transmission.

**Treatment.** In order to satisfy the patient’s aesthetic needs, rehabilitation was performed on the anterior teeth. A direct restoration technique was used with composite resin materials. The main goal in restoring functional rehabilitation was to reconstruct the permanent molars as there had been extensive wear (Figs. 2a, b). Before going ahead with the reconstruction the vertical height posteriorly and the overjet had to be corrected. This was achieved by using a Frankel type orthodontic functional device.

After a 10 month period of orthodontic therapy an adequate molar extrusion was achieved that allowed the next restoration phase. In the first part the permanent molars were reconstructed. The technique used was as follows: the area to be operated was isolated with rubber dam and the occlusal surface was prepared with a high speed bur so as to attain a retention shape. Orthophosphoric acid (10%) was applied for 30 seconds and the teeth were rinsed and dried. A layered polymerisation technique with composite resins was used to achieve the final restorations (Figs. 3, 4a, b).

Three month follow-ups were arranged to monitor the stability of the therapeutic results over time. The patient was encouraged to use the oral hygiene procedures laid down for her at home. After two years the composite resins were still in place and oral hygiene was satisfactory.

![Fig. 1 - Photograph showing: a) anterior aspect of affected incisors; b) increased incisor overjet.](image1)

![Fig. 2 - Photograph showing affected posterior teeth: a) mandible; b) maxilla.](image2)
Discussion

AI hypoplastic type is different from other forms of AI because of radiographic, clinical, histologic, and biochemical characteristics. The enamel ultrastructure shows a reduction of enamel thickness combined with a loss of prism structure, especially on the surface [Ooya et al., 1988; Seymen and Kiziltan, 2002]. The calcium concentration is very low in the AI enamel layers directly adjacent to the amelo-dentinal junction and shows a steeper concentration gradient. Histopathological analysis shows pre-dentine areas with irregular canaliculi between normal dentine and internal resorption areas in the pulp tissues [Wright et al., 1991]. The calcium concentration is very low in the AI enamel layers directly adjacent to the amelo-dentinal junction and shows a steeper concentration gradient. Histopathological analysis shows pre-dentine areas with irregular canaliculi between normal dentine and internal resorption areas in the pulp tissues [Wright et al., 1991]. Studies of AI using electron microscope show that calcified bodies located in the gingival tissues appear to be composed, in part, of a dense enamel-like substance and also of a tissue with features of afibrillar cementum [Ooya et al., 1988].

Therapeutic approach. The therapeutic approach to patients with AI is determined by the clinical severity of the lesions and the need for an aesthetic improvement [Gibbard, 1974; Seow, 1993; Rosenblum, 1999; van Waes and Stockli, 2001]. Above all, it is necessary to set up a protocol of primary prevention based on domestic and professional oral hygiene checked every three months using a topical fluoride prophylaxis and fissure sealant program. Motivation of the child and its parents to oral hygiene and basic prevention is very important. The paediatric dentist must carry out frequent periodic controls to ensure that the prescribed recommended procedures are being correctly and constantly applied. The use of fluoride gels can be of indispensable assistance for those patients with the highest caries risk.

A fundamental preventive intervention should consist of the sealing of grooves and pits. With patients with normal dental development the intervention is effected primarily on the first permanent molars. With patients at high risk, for example those affected by AI, it should be undertaken also for primary teeth. At a later stage of treatment, aesthetic considerations may be dealt with by way of a restorative approach using reconstruction of the anterior teeth. This should also be with functional rehabilitation through reconstruction of the occlusal surfaces of the posterior molars with composite resin materials.
[Seow and Amaratunge, 1998; Dias et al., 2001; Soares et al., 2002].

With adults, when the structure of the dentine is normal, preparation may be effected following the same principles as for healthy teeth for the fitting of crowns. But, as coronal pulps retract more rapidly than with normal teeth, due to a thin and incomplete enamel covering, coronal preparation may be carried out on younger patients. In some cases of hypomaturation and hypoplasia, restoration with facings may, however, be a more preservative alternative for aesthetic problems of anterior teeth.

However, it should be emphasised that the orthodontic treatment carried out on patients affected with AI is often an indispensable phase in functional rehabilitation [Cooke and Neesome, 1990; Hoppenreijs et al., 1998; Rosemblum, 1999]. Specifically, the use of active functional orthodontic devices may be validly employed during the preliminary phase of treatment. With devices such as the Frankel type it is possible to obtain an orthodontic extrusion of the lateral-posterior sectors. This is not only essential in order to then carry out correct preservative preparations, but above all a remodelling of the dental arch at the anterior segments, so allowing an improved control of the interdental spaces and, therefore, an improved stability of the aesthetic reconstruction over time.

**Conclusion**

The importance of an early ortho-paedodontic intervention in individuals with amelogenesis imperfecta is emphasised. The correct planning of treatment timing should be for a regular program of professional oral hygiene, aesthetic and functional re-establishment of the dentition and the correct development of teeth.

**References**


