

Case Report

Management of Amelogenesis Imperfecta: Report of Three Cases

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Abstract: Amelogenesis Imperfecta (AI) is a genetic disease affecting primary and permanent tooth enamel. The incidence varies between 1:700 to 1:4000. The clinical findings include enamel defects, tooth sensitivity, poor dental aesthetics, reduced vertical dimension, dentin dysplasia, and pulpal calcification. Effective treatment planning should incorporate numerous factors such as the patient's age, disease type and severity, and general oral condition. Over time, severe tissue destruction may occur, and therefore, it is important to begin treatment as early as possible. The present case report describes patients diagnosed with three types of AI: hypoplastic, hypocalcified, and hypomaturation.

Keywords: Amelogenesis Imperfecta, Hypoplastic, Hypocalcified, Hypomaturation

INTRODUCTION

Amelogenesis Imperfecta (AI) is a heterogeneous hereditary anomaly that disrupts enamel tissue development and emergence, independent of any systemic disorder [1-3]. This enamel anomaly occurs in all or part of the tooth and affects both permanent and primary dentition [2-4]. The disease is inherited in an autosomal dominant, autosomal recessive, or X-linked pattern, but autosomal-dominant is the most common mode of inheritance [6, 7]. There several classification systems for the disorder were originally devised in the 1940s [8-10] but Aldred *et al.* [2] have since reclassified AI into four groups: genetic, molecular origin, biochemical, and phenotypic. They further subclassified the clinical presentations into hypoplastic, hypocalcified, hypomaturation, and hypomaturation-hypoplastic with taurodontism categories. In hypoplastic AI, the decreased enamel thickness is caused by a defect in matrix formation; the enamel composition is still hard and varnished, but the teeth appear malformed. Hypocalcified AI is caused by an enamel calcification. The enamel has a normal thickness but weak structure and is visibly chalky and opaque. In addition, the enamel is friable and easily removed from the underlying dentin, and radiographically, the enamel appears less radiopaque than dentine. In the hypomaturation type, enamel maturation time is defective, which results in a softer enamel with a mottled, notched appearance and opaque white, yellow-brown, or red-brown color. Radiographically, it is similarly radiodense to dentin.

Hypomature-hypoplastic with taurodontism AI is both hypoplastic and hypomature in appearance, which is caused by taurodontism [1, 11-13]. AI-affected patients experience tooth eruption abnormalities, tooth sensitivity, poor aesthetics, chewing difficulties, high caries risk, and vertical dimension losses [14]. In addition to these congenital defects, open occlusal relationships, pulpal calcifications, dentin dysplasias, hypercementosis, root and crown resorption, root malformations, and taurodontism may also occur in patients diagnosed with AI [15-18].

CASE REPORT 1

A 12-year-old boy was referred to the Department of Pediatric Dentistry complaining of aesthetic problems. Extraoral examination revealed no facial asymmetry or TMJ disorders. On oral examination, the enamel was discolored yellow-brown on all teeth (Figure 1). The enamel itself appeared normal, but was indented (Figure 2). After clinical and radiologic examination, the patient was diagnosed with hypomaturation type AI. Dental caries were detected in teeth 36 and 46 (Figure 3). The caries were restored, fluoride treatment was applied, and the condition was followed-up periodically.



Fig. 1: Intraoral appearance of Case 1



Fig. 4: Intraoral appearance of Case 2



Fig. 2: The enamel of the teeth appeared normal, but indented



Fig. 5: Composite restoration was applied to the Case 2's incisor teeth



Fig. 3: Dental caries in teeth 36 and 46

CASE REPORT 2

A 7-year-old girl was referred to the Department of Pediatric Dentistry for aesthetic complains. Extraoral examination of the patient was normal. However, while her oral examination showed a normal enamel structure, it also revealed thinned, conical and cylindrical shaped crowns with no contact area (Figure 4). Hypoplastic type AI was diagnosed after clinical and radiologic patient examination. A composite restoration was applied to the patient's incisor teeth (Figure 5).

CASE REPORT 3

A 6-year-old boy and his family were referred to the Department of Pediatric Dentistry complaining of tooth sensitivity. His extraoral examination did not reveal any abnormality. An oral examination showed yellow-brown enamel discoloration and attrition of the incisal and occlusal surfaces (Figure 6). On the basis of clinical and radiologic examination (Figure 7), the patient was diagnosed with hypocalcified type AI, and his sensitive teeth were restored. The patient was educated on proper oral hygiene and was followed up periodically.



Fig. 6: Intraoral appearance of Case 3



Fig. 7: Panoramic radiograph of Case 3

DISCUSSION

The abnormal aesthetic appearance, function, occlusion, phonation, and other negative gingival effects of AI require a different approach from traditional dental treatment. Aesthetic abnormalities such as tooth sensitivity, discoloration, and abnormal texture cause psychological and functional problems for the patient. Treatment planning must focus on early diagnosis, pain management, prevention, stabilization, restoration of defects, and regular long-term management [13]. The treatment plan should also accommodate factors including the patient's age, socio-economic status, disease type and severity, and overall oral condition [18]. Affected patients are typically treated with hard porcelain crowns, composite restorations, stainless steel crowns, laminate applications, or overdenture applications [2, 8]. The therapeutic goals for these patients should focus on recovering aesthetic appearance and functional phonation, as well as preserving gingival health.

Treatment of hypoplastic and hypocalcified type AI is comparatively different [19-21]. Sundell [19] reported that teeth were treated with prosthetic restoration in hypocalcified AI, while hypoplastic type teeth were treated with composite restoration.

In the first case, the patient's decayed tooth was restored, fluoride treatment was applied, and the patient was reexamined periodically. Prosthetic restoration has been postponed until dentition growth and development are complete. In the second case, the patient regained aesthetic function and phonation once her tooth was restored with composite. She was educated on proper oral hygiene and followed-up periodically. In the third case, the patient had difficulty once the permanent teeth began erupting. Depending on the sensitivity and fragmentation present, there may be obstacles to providing good oral hygiene and tooth restoration. After treating the patient's tooth sensitivity, oral hygiene education was provided, and the patient was followed-up periodically. Malocclusion may develop in affected children, and therefore treatment

should incorporate both the pediatric and restorative dentistry in a multidisciplinary approach [13]. Depending on the specific calcification, clinical changes, and malocclusion present, the pediatric dentist, orthodontist, and oral maxillofacial surgeon may be involved in care. In these patients, aesthetic and functional problems must be resolved, and regaining the patient's psychosocial trust should be prioritized. Tissue destruction increases in affected patients, and therefore prevention of tooth loss is important in the treatment plan.

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