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Clinical Diagnosis and Oral Rehabilitation of a Patient with Amelogenesis imperfecta: A Case Report

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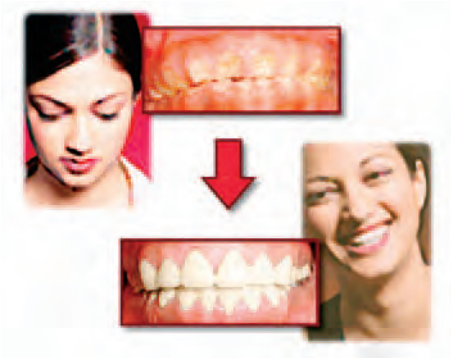

Abstract

Aim: This clinical report describes the oral rehabilitation of a young female patient diagnosed with the hypocalcified, autosomal recessive type of Amelogenesis imperfecta (AI). A brief discussion on diagnosis of AI is also included.

Background: AI has been defined as a group of hereditary enamel defects not associated with evidence of systemic disease. It can be characterized by enamel hypoplasia and/or hypomaturation or hypocalcification of the existing teeth. Restoration for patients with this condition should be oriented toward the functional and esthetic rehabilitation and the protection of these teeth.

Report: A 31-year-old female patient presented with concerns including extreme sensitivity; dissatisfaction with size, shape, and shade of teeth; and poor masticatory efficiency. She was very conscious about the appearance of her teeth and reported that her primary dentition was affected in the same manner. The specific objectives of this treatment were to eliminate tooth sensitivity, enhance esthetics, and restore masticatory function. Treatment included crown lengthening procedures and placement of anterior and posterior metal-ceramic crowns. A 12-month follow-up with clinical and radiographic examinations revealed no evidence of any untoward effects of the treatment on the restored teeth or their supporting structures.

Summary: Management of a patient with AI is a challenge for the clinician. The treatment options vary considerably depending on several factors such as the age of the patient, socio-economic status, periodontal

condition, loss of tooth structure, severity of the disorder, and, most importantly, the patient's cooperation. The clinician has to consider the long-term prognosis of the treatment outcome. This clinical report describes the fabrication of metal ceramic and all metal crowns for the restoration of severely worn teeth in a patient with AI which requires meticulous maintenance of oral hygiene and patient co-operation.

Keywords: Amelogenesis imperfecta, AI, hereditary enamel defects, clinical diagnosis, full-mouth rehabilitation

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Introduction

Amelogenesis imperfecta (AI) has been defined as a complex group of hereditary enamel defects not associated with evidence of systemic disease[1,2] affecting both primary and permanent dentitions.[1] It is a rare enamel mineralization defect described by Spokes[3] in 1890 as "hereditary brown teeth" with a reported incidence of 1:14,000.[4]

Phenotypically AI is categorized into four broad groups: hypoplastic, hypomaturation, hypocalcified, and a hypomaturation-hypoplastic variety. Fifteen subtypes of AI exist phenotypically and based on modes of inheritance. This classification has been proposed by Witkop[5] (Table 1).

The types are characterized as follows:

- **Type-I:** Lesions may appear as pin-point to pinhead sized pits scattered across the surfaces of teeth. The distribution of lesions may be generalized or localized, and the alteration of the enamel is a result of inadequate deposition of enamel matrix.
- **Type-II:** Also known as the hypomaturation type is associated with abnormalities in the maturation stages of enamel formation resulting in the enamel being opaque and chalky in appearance. The enamel layer is normal in thickness but softer than normal and can be easily detached from the underlying dentin.
- **Type-III:** The teeth have enamel that is insufficiently mineralized and clinically appears as severely worn teeth. It results from detachment of the enamel from dentin within a short period after tooth eruption. Teeth are very sensitive to thermal changes and appear dark brown in color.
- **Type-IV:** AI exhibits enamel hypoplasia in combination with hypomaturation. This variety is associated with taurodontism. The most common form of AI is the autosomal dominant hypocalcified type, followed by hypomaturation, and the hypoplastic type.[6]

Other associated findings in patients with AI include delayed eruption of teeth, taurodontism, congenitally missing teeth, crown and root resorption, and pulp calcification.[7] Radiographically the density of enamel layer is lower than normal enamel. Hypoplastic enamel shows great variation in density and it may be difficult to distinguish it radiographically from underlying dentin.

AI is caused by mutations in a variety of genes that are critical for normal enamel formation. A total of about five genes [AMELX, ENAM, KLK4, MMP20, and DLX3][8,9] are known to be involved in enamel formation. Mutations of the amelogenin gene (AMELX) cause X-linked AI, while mutations of the enamelin (ENAM) gene causes autosomal inherited forms of AI. Other genes like Kallikrein – 4 (KLK4), MMP–20, and DLX3 genes contribute to the etiologies of some other varieties of AI which is still under investigation.

Various treatment methods or strategies were initially instituted for AI patients such as the extraction of the compromised teeth and placement of a removable prosthesis or implant supported fixed or removable prosthesis.[10] However, these procedures are very invasive and have greater incidence of complications. Numerous treatment modalities have been described for rehabilitation of patients with AI.[10-17] Rehabilitation of patients with AI requires meticulous oral hygiene maintenance and patient cooperation.

This rare dental abnormality poses a major restorative challenge for the dentist. Using

Table 1. Classification of AI according to Witkop[5] (1989).

Type I	**Hypoplastic**
IA	Hypoplastic, pitted autosomal dominant
IB	Hypoplastic, local autosomal dominant
IC	Hypoplastic, local autosomal recessive
ID	Hypoplastic, smooth autosomal dominant
IE	Hypoplastic, smooth X-linked dominant
IF	Hypoplastic, rough autosomal dominant
IG	Enamel agenesis, autosomal recessive
Type II	**Hypomaturation**
IIA	Hypomaturation, pigmented autosomal recessive
IIB	Hypomaturation, X-linked recessive
IIC	Snow-capped teeth, autosomal dominant?
Type III	**Hypocalcified**
IIIA	Autosomal dominant
IIIB	Autosomal recessive
Type IV	**Hypomaturation-hypoplastic with Taurodontism**
IVA	Hypomaturation-hypoplastic with taurodontism, autosomal dominant
IVB	Hypoplastic-hypomaturation with taurodontism, autosomal dominant

conservative techniques desirable esthetics can be achieved, the teeth and supporting structures preserved, and a harmonious relationship created between the occlusion and temporomandibular articulation.

Case Report

Diagnosis

A 31-year-old female patient presented with considerable dental sensitivity and wear of her teeth. Her primary concerns included extreme sensitivity; dissatisfaction with size, shape, and shade of teeth; and poor masticatory efficiency. She was very conscious about the appearance of her teeth and reported that her primary dentition was affected in same manner. A detailed medical history, dental history, and social history was obtained but was noncontributory. The patient was questioned further about the presence of similar abnormalities in her family. She stated her grandfather had a similar defect in his teeth.

An extraoral examination revealed no abnormalities. Intraoral examination revealed a full complement of the permanent dentition.

The incisal aspects of maxillary and mandibular anteriors were completely worn away exposing the pulp chambers (Figure 1).

The occlusal aspects of all the posterior teeth were also severely worn, however, the cervical and proximal enamel was found to be normal. The crowns were short and thin with the tooth surfaces being rough and dull. Deep carious lesions were seen in the right and left mandibular molars. The attrition of the molars resulted in a decrease of the vertical dimension of occlusion. The interocclusal distance at physiologic rest position was 7.3 mm, and the centric occlusion position was coincident with the maximum intercuspal position. The gingival status was found to be good and well maintained. The oral hygiene of the patient was satisfactory.

A panoramic radiographic examination of the teeth revealed generalized defective enamel on all the teeth (Figure 2). The enamel of the teeth appeared to have the same radiodensity as dentin and the morphology of the roots were normal. The pulp chambers were normal with no evidence of calcification. The cementum, lamina dura, and bony trabeculations were within normal limits.

The hypocalcified variety of AI is characterized by enamel that is insufficiently mineralized. It is soft and easily lost over a short period if time following eruption. The current patient presented with similar clinical features such as the enamel being chipped off and the teeth severely worn exposing the pulps of several teeth. Hence, hypocalcified variety of AI was considered. A positive history of a similar problem with the patient's grandfather prompted an investigation into possible occurrences in other members of the family. Observations indicate the disease had skipped a generation in between the grandfather and the patient. The patient's siblings did not express the disease, suggesting a recessive inheritance (Figure 3). Based on the patient history and clinical examination a diagnosis of AI (hypocalcified autosomal recessive type) was made.

Treatment

To satisfy the patient's primary concerns, a treatment plan was developed to include an oral prophylaxis and oral hygiene instructions followed by lengthening of the clinical crowns

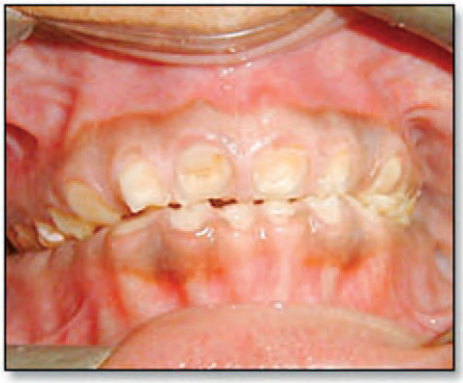

Figure 1. Pretreatment photograph of the patient with AI showing complete attrition of teeth.

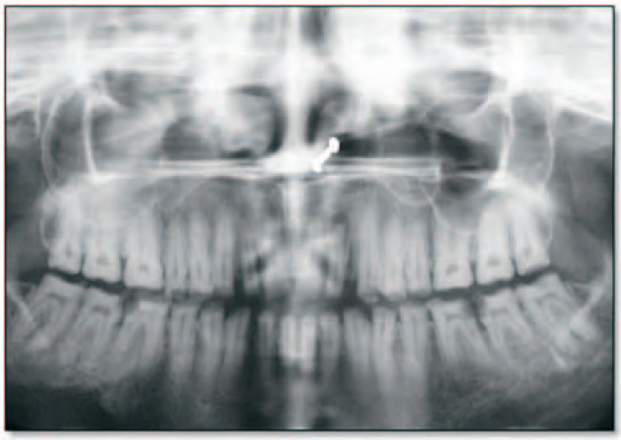

Figure 2. Panoramic radiograph showing generalized defective enamel in all teeth with its radiodensity being the same as the dentin. The morphology of roots is normal. A pseudocyst is apparent in the right maxillary sinus.

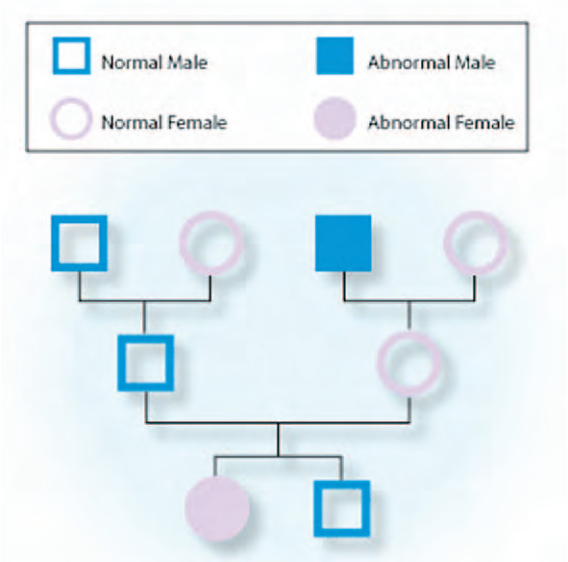

Figure 3. Pedigree chart of AI (hypocalcified autosomal recessive variety).

of all teeth followed by placement of metal-ceramic crowns. The patient was prescribed 0.12% chlorohexidine gluconate mouth rinse twice daily. Since the heights of the crowns of the maxillary and mandibular teeth were inadequate for the fabrication of the prosthesis, an apically positioned flap was planned as a part of the crown lengthening procedure with consideration for biologic width dimensions. The surgical site was allowed to heal for three months. Finally, increase of crown height by approximately 2 mm was achieved. Figures 4 and 5 show the pre-treatment and post-treatment photographs of the lower right quadrant.

Caries excavation was done for all carious teeth. Endodontic therapy was carried out on all teeth except the maxillary posterior teeth to address the multiple pulp exposures caused by the severe attrition.

Maxillary and mandibular complete-arch impressions were made using Hydrogum Soft irreversible hydrocolloid (Thixotropic, Zhermach, Italy) impression material. Diagnostic casts were fabricated from Type-III dental stone (Pankaj Industries, Mumbai, India) and mounted on a Whipmix semiadjustable articulator using a face bow transfer. Bite registration using Type II modeling wax (Hyderabad Dental Products, Hyderabad, India) was made at an increased vertical dimension of 5 mm with 3 mm of freeway space. Using these records a splint was fabricated with heat-cured polymethyl methacrylate acrylic resin (DPI-Heat cure, Dental products of India Ltd, Mumbai, India). This splint was used to remove muscle engram (muscle deprogramming). The patient used the splint for three months while the teeth were restored with temporary restorations without any complications.

After completion of endodontic therapy, the maxillary anterior teeth were prepared with post spaces for cast post cores and for prefabricated posts for the mandibular anterior teeth. Composite core build-ups (Clearfil Photocore, Kuraray Dental, Japan) were fabricated for the premolars and the right first molar in order to increase the crown height. (Figure 6)

In order to avoid trauma to the gingival sulcus a thin retraction cord was inserted into the

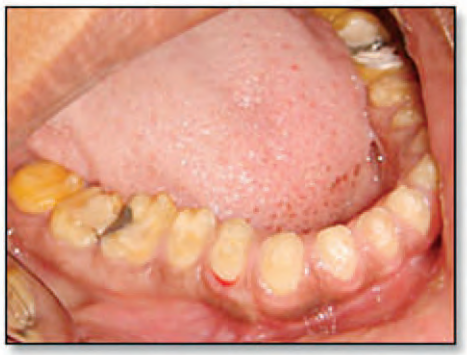

Figure 4. Lateral view of the mandibular teeth showing inadequate crown height for the fabrication of a prosthesis.

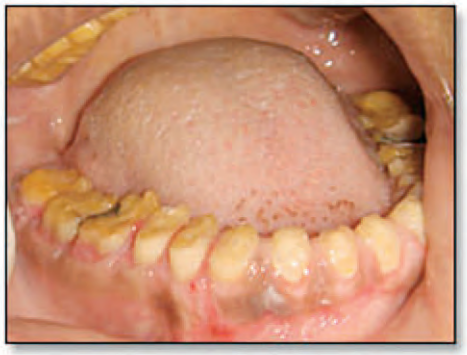

Figure 5. Post treatment photograph of the patient showing surgically lengthened crowns with an increase in the crown height by approximately 2 mm.

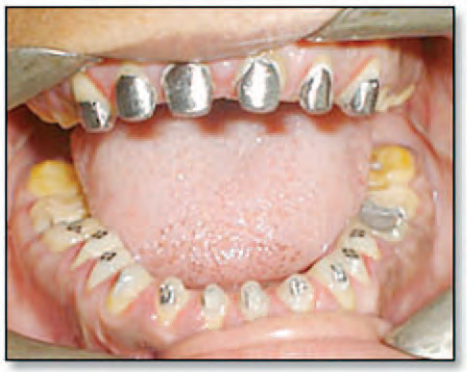

Figure 6. Anterior view of the teeth showing cast post and core in maxillary anteriors and prefabricated posts and composite build up in mandibular anteriors.

sulcus prior to preparation. Crown preparations were done for porcelain-fused-to-metal (PFM) restorations for the maxillary and mandibular anteriors, premolars, and maxillary first molars; on the remaining teeth all-metal restorations were used. Gingival displacement prior to impression making was done using a non-hemostatic gingival magic foam cord retraction system (Ultradent, South Jordan, UT, USA).

Impressions were made with addition polyvinyl siloxane material (Reprosil, Dentsply/Caulk; Milford, DE, USA) using the putty wash technique (Panasil Putty Soft, Dentsply Kettenbach, Germany). Full-mouth, heat-cured provisional restorations (Figure 7) were fabricated at the desired vertical dimension (with 3 mm freeway space) using methyl methacrylate acrylic resin. The provisional restorations were temporarily cemented using Provicol, eugenol free $Ca(OH)_2$ cement (Voco, Cuxhaven, Germany).

The patient wore the provisional restorations at the newly established occlusal vertical dimension for three months. Final impressions were made using the putty-wash technique and casts were prepared using Type IV stone in conjunction with the Pindex system (Confident, Bangalore, India) to create removable dies. The working casts were mounted onto the Hanau Widevue semiadjustable articulator (Waterpik, Ft Collins, CO, USA) using Type I rigid tray material with the interocclusal records (Take 1, Kerr, Romulus, MI, USA). Wax patterns were made using inlay wax (Harward, Harward, Germany) and then casting was done using Metal ceramic alloy (Remanium CSe, Dentaurum J.P. Winkelstroeter KG, Ispringen, Germany). During the cast metal try-in marginal fit and passivity were evaluated (Figure 8).

The appropriate shade was then selected using the VITA shade guide (Vita Zahnfabrik, Badsackingen, Germany) and porcelain firing was done.

The PFM restorations for the maxillary and mandibular anteriors, premolars, and maxillary first molars were cemented temporarily with Provicol for two weeks then permanently cemented with GIC Fugi II cement (GC, Tokyo, Japan) (Figure 9).

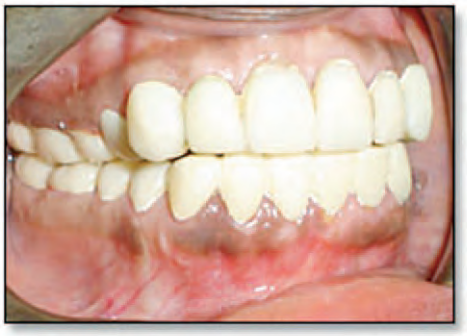

Figure 7. Photograph showing full mouth heat cure processed provisional restoration which was fabricated at the desired vertical dimension.

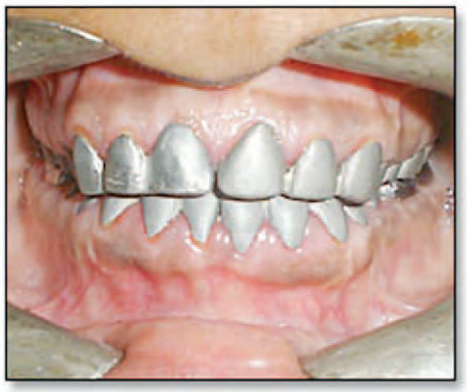

Figure 8. Photograph showing the cast metal try-in for evaluation of marginal fit and passivity.

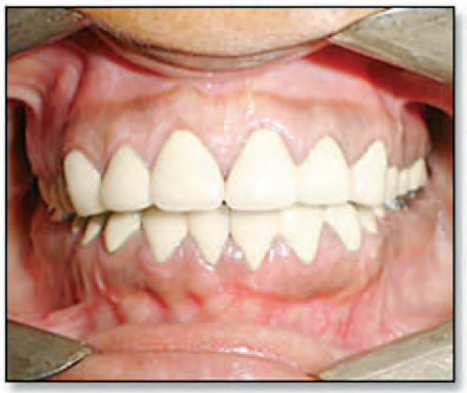

Figure 9. Photograph showing anterior view of the rehabilitated dentition in occlusion, one year after treatment.

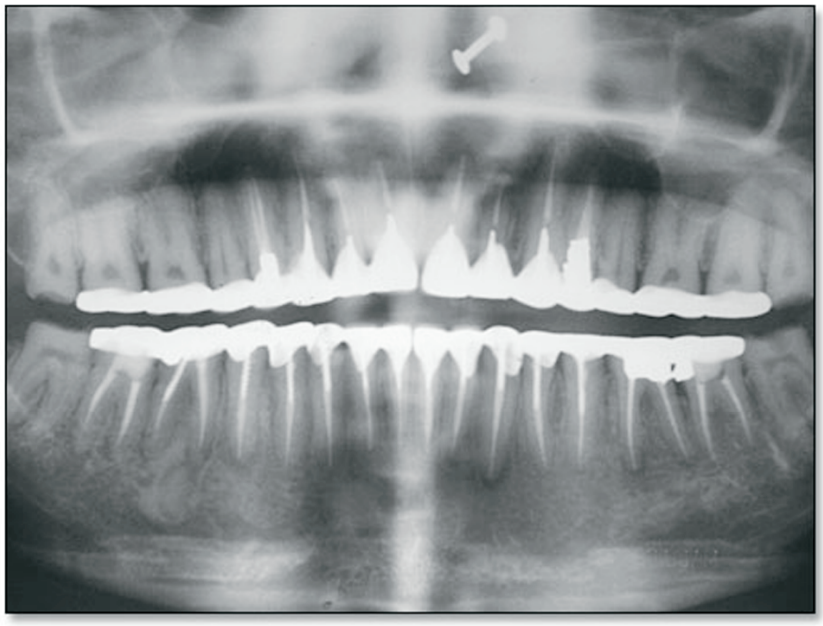

Figure 10. A post-operative panoramic radiograph of the patient's dentition one year after treatment.

Since there was no concern regarding esthetics, full cast metal restorations were fabricated for the remaining teeth. Oral hygiene instructions were given again.

The crowns were splinted together with wide embrasure spaces for maintenance to provide adequate retention and resistance form. The disadvantages of splinting the teeth include the need for frequent follow up visits and meticulous oral hygiene measures.

The patient was satisfied with the treatment outcome and is following a strict three month recall program. The patient's dental sensitivity disappeared and normal eating habits were established. Figure 10 is a panoramic radiograph taken at the one year recall visit which showed no evidence of any disorder associated with the restored teeth or their supporting structures. The psychological change which was seen in the patient was appreciable.

Discussion

Based on the clinical presentation and strong family history a diagnosis of AI (hypocalcified, autosomal recessive type) was made. An accurate diagnosis is important clinically for several reasons. First, one has to exclude the presence of systemic diseases that may show generalized enamel hypoplasia. Secondly, an accurate diagnosis enables genetic counseling which is often sought by affected families. Finally, an accurate diagnosis helps in recognition of the condition so preventive measures can be provided early.

According to Seow[2] the primary clinical problems of AI are esthetics, dental sensitivity, and loss of vertical dimension. These patients are highly susceptible to dental caries, gingival inflammation, as well as an anterior and posterior open bite.

Treating the patient with AI is important for functional and psychosocial reasons. Some patients need only oral hygiene instructions, while others need extensive dental treatment. Historically treatment of such patients had included extractions and fabrications of complete dentures, however, these options are psychologically displeasing to the patient.

The following are general treatment steps suggested for patients with AI:

Preventive and Initial Phases:

- Oral hygiene instructions and oral prophylaxis.
- Chlorhexidine mouth rinses.
- Topical fluoride application.
- Control of dentinal hypersensitivity using desensitizing agents.

- Extraction of teeth which have a poor prognosis.
- The initial provisional stage of treatment should be performed as soon as AI is diagnosed when the patient is cooperative.

Restorative Phase:

- Establish a favorable occlusal vertical dimension using a provisional occlusal splint.
- Composite build up of the teeth with severe loss of tooth structure.
- Fabricate thin gold crowns for posterior teeth.
- Lengthen the crowns of the worn teeth.
- Fabricate metal-ceramic crowns, all ceramic crowns, or porcelain veneers if the enamel is suitable for bonding for teeth where esthetics is a concern.

Maintenance Phase:

- Monitor oral hygiene, periodontal, and pulpal status.

Because of the recent advances in the field of esthetics and prosthetic dentistry, it is possible to restore the function and esthetics to an acceptable level in severe AI cases.

Summary

Management of a patient with AI is a challenge for the clinician. The treatment options vary considerably depending on several factors such as the age of the patient, socio-economic status, periodontal condition, loss of tooth structure, severity of the disorder, and, most importantly, the patient's cooperation. The clinician has to consider the long-term prognosis of the treatment outcome. This clinical report describes the fabrication of metal ceramic and all metal crowns for the restoration of severely worn teeth in a patient with AI which requires meticulous maintenance of oral hygiene and patient co-operation.

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