

Amelogenesis imperfecta: a case report

Edward Huen-Tai Ho ^{*}, BDS, MCLinDent, LDSRCS, FDSRCS, FRACDS, MRDRCS & RCPS

ABSTRACT Amelogenesis imperfecta is a hereditary condition where enamel formation is disturbed resulting in defects in mineralization or matrix formation. Restoration of the dentition poses great difficulties especially when all the teeth are severely affected. Treatment aims to relieve pain or tooth sensitivity, to preserve as much tooth tissue as possible while preventing further tooth loss, to maintain masticatory function, and, last but not least, to improve the appearance as this has great psychological impact on the patient's confidence. This article describes treatment of a girl who presented with a severe form of amelogenesis imperfecta.

Introduction

Amelogenesis imperfecta (AI) is a collective term for a number of conditions resulting from abnormal enamel formation. Many cases are inherited, either as autosomal dominant, autosomal recessive or as an X-linked trait. There have been a number of classifications, based primarily on the phenotype ¹⁻³. However, a recent classification based on the molecular basis for the observed enamel phenotype may help clarify the commonly seen overlap in phenotypes. According to this classification, AI is a group of conditions, genetic in origin, which affect the structure and clinical appearance of the enamel of all, or nearly all the teeth, and which may be associated with morphologic or biochemical changes elsewhere in the body ⁴. This enamel anomaly affects both primary and permanent dentition ^{4,7}. The incidence of AI has been reported as varying between 1:700 and 1:16 000, depending on the diagnostic criteria and the population studied ^{2,7-9}. The appearance depends on the type of AI, varying from the mild hypomature 'snow-capped' enamel to the more severe hereditary hypoplasia with thin, hard enamel which has a yellow-brown appearance. Dental problems, which depend on the severity of the condition, include sensitive teeth and poor appearance due to tooth loss and staining. If tooth tissue loss is severe there is vertical loss resulting in reduced masticatory function and

poor appearance. Pulpal involvement may occur in severe cases, and because the roughness makes cleaning more difficult, gingivitis and periodontitis may develop. As AI affects both primary and permanent dentition, prevention of future dental problems must start from the beginning, by educating the parents and reviewing the patient regularly. Attention must also be paid to the psychological health of the patient ¹⁰.

Case report

The patient was a healthy 17-year-old Nigerian student (Figure 1) referred to the author's hospital by a general dental practitioner in June 2000. She was complaining of poor appearance, sensitive teeth, and was unable to eat properly. She had emigrated from Nigeria a few years previously. She had a severe form of AI, with absence of all the enamel when first seen (Figure 2). Her primary teeth had also been affected but she was not sure to what extent. Those teeth present were heavily stained, had no deep carious lesions and the exposed dentine was relatively softer than the normal dentine. The teeth were vital, firm, and not tender to percussion. Only some teeth were sensitive to cold. These were vital and radiographs showed narrow pulps and root canals. The periodontal tissues were healthy. Wisdom teeth (38 absent) could be felt behind the second molars.

Her elder brother also suffered from the same type of problem but it did not affect her younger sister and brother. The elder brother's dental health was even worse than the patient's and he had been essentially treated with extraction and the provision of dentures.

Treatment objectives for this patient were (1) prevention of caries and gingivitis; (2) improvement of esthetics;

* Private practice as Specialist in Prosthodontics

Correspondence to:
Dr. Huen-Tai Ho
Room 1005B, 10/F, Argyle Centre, Phase 1, 688 Nathan Road,
Kowloon, Hong Kong
Tel : (852) 2397 1628, 2397 1629
Fax : (852) 2380 8968
e-mail : ehtho@netvigator.com



Figure 1 Before treatment

(3) prevention of further deterioration of the remaining dentition; (4) patient education and motivation; and (5) management of sensitive teeth. There were several treatment options: provisional crowns, composite restorations or occlusal splint at a predetermined vertical dimension and then definitive crowns when the patient adapted to the new vertical dimension. The option involving provisional crowns was selected because all the teeth urgently required protection and their appearance was poor.

As such treatment entails a very long time commitment, patient cooperation is essential if these objectives are to be achieved.

Treatment plan

Treatment was divided into three phases:

Phase 1

1. Oral hygiene instruction and dietary advice.
2. Mounting of study models and evaluation of occlusion.
3. Diagnostic waxing up of upper and lower anterior teeth.



Figure 2 (a) Frontal view and (b) occlusal view of upper teeth; (c) occlusal view of lower teeth

4. Discussion of treatment options.
5. Blow-down matrices for upper and lower temporary crowns, the re-organized jaw relationship raising the incisal pin by 4 mm.
6. Teeth 13 to 23, and 33 to 43 prepared and impressions taken for provisional crowns. Temporary crown cementation at pre-determined vertical dimension. Posterior teeth build-up with composite to maintain new occlusal relationship.



Figure 3 Treatment completed

7. Provisional acrylic crown cementation then regular patient review until new occlusal scheme problem-free.
8. Surgical extraction of all three wisdom teeth arranged (due to their potential for causing periodontal problems).

Phase 2

1. Three months after adaptation to the new occlusal vertical dimension (OVD), the new occlusal relationship was recorded using new study models and face-bow records.
2. Diagnostic waxing up of the upper and lower posterior teeth was done prior to the making of upper and lower blow-down matrices.
3. Upper posterior teeth were prepared for metal ceramic crowns.
4. This was followed by provision of lower posterior metal ceramic crowns.
5. Lower anterior teeth were replaced with Procera crowns.
6. Upper anterior teeth were replaced with Procera crowns. Crowns were made using the custom-moulded incisal table (from the upper anterior provisional crowns) to determine the palatal contours (Figure 3).

Phase 3

1. Oral hygiene reinforcement.
2. Dietary advice.
3. Regular dental checkups for caries and periodontal diseases.
4. Regular review of occlusion (Figure 4).

Discussion

Amelogenesis imperfecta is an inherited disorder, ranging from a defect in enamel formation to co-



Figure 4 One-year review

existence of other medical disorders¹¹⁻¹³. As both the primary and permanent dentition are affected, preventive measures should be started, even before the teeth erupt.

In this case, the patient presented with severe tooth wear due to inherited abnormal enamel development. The poor appearance was due not only to the innate color of the teeth, but also to increased staining, chipping and attrition of the teeth, and spacing between the teeth. Discomfort was due to wide areas of exposed dentine. There was no pulpal involvement as treatment was initiated before further damage was done.

Treatment options for such a case include the following:

1. Provisional crowns made to the length and shape required for permanent anterior crowns. Posterior teeth restored with composite to the new vertical dimension. Definitive crowns made when the patient has adapted to the new vertical dimension. Extraction of wisdom teeth.
2. Composite restorations of all except the wisdom teeth

to the new vertical dimension. Definitive crowns when the patient has adapted to the new dimension. Extraction of the wisdom teeth.

3. Occlusal splint at the new vertical dimension, then definitive crowns made when the patient has adapted to the new occlusal relationship.

The first option was selected because all her teeth urgently required protection and the appearance was poor. Provisional crowns also provided valuable information about future restorations. As treatment required a very long time commitment, cooperation was stressed right from the beginning.

Re-organization of the occlusion was undertaken because all the teeth required restoration and the OVD had to be altered¹⁴. Evaluation of the patient's ability to adapt to the new OVD and establishment of a stable retruded contact position (RCP) must be acquired prior to the definitive treatment. Either an occlusal splint or provisional restorations like provisional crowns and intermediate composite can be used. The author preferred the latter option as the need to convert the splint into temporary restorations could be omitted; at the same time the teeth were protected and appearance improved. In addition, appearance and phonetics could be evaluated during this trial period. Stabilization of the jaw with anterior provisional crowns and posterior composite to the new vertical dimension were carried out initially. Excursive interferences were adjusted and definitive treatment could be started when a stable RCP was acquired with no change occurring between two successive appointments and the patient adapting to the new OVD. The time needed for this ranges from a couple of weeks to several months¹⁵. In order to analyze the appearance and the patient's ability to adapt to the new OVD, definitive treatment should not begin for at least one more month^{16,17}. In this case, adaptation took longer than 1 month. Owing to school examinations, definitive treatment was initiated 2 months later. After the anterior guidance was determined and copied, the posterior teeth were definitively restored. Shallow anterior guidance was planned to reduce stresses on the incisors, resulting in shallower cusps in the posterior crowns.

Removal of caries had to be done very carefully as the defective dentine was softer than normal dentine. Because the defective dentine involved a large area and in many areas was quite subgingival, there was a problem with obtaining all the preparations in one impression. Electrosurgery was considered initially, but later abandoned

after minimal success and because most of the teeth were involved. It would be very traumatic and uncomfortable for the patient. A double-cord technique was used but the result was more successful with the anterior teeth. The problem of getting a good impression of all the posterior teeth was solved by the pick-up copings. Good dies were inserted into the corresponding copings so all the dies could be assembled together. This was found to be very helpful as fit of crown margins could be evaluated easily and all the crowns could be assembled together.

Metal ceramic crowns for 27 and 37 had to be changed into full gold crowns because the patient did not return after the composite on 37 was debonded. Although the patient was informed at the beginning of treatment of the effect of losing the composite or provisional crowns, she did not return when the composite on 37 was debonded. Because the space available made it possible to use gold crowns and the patient accepted gold crowns as alternative treatment, full gold crowns were made for 27 and 37.

The morphology of the anterior crowns was constructed according to the racial characteristics of the patient, closing up the upper and lower midline diastemata and copying the provisional crowns as requested. Owing to the highly discolored dentine a more opaque ceramic material had to be used. Although the crowns could have been improved further, she was very happy to accept them; thus no further alteration was needed.

Construction of 28 crowns demanded very strong laboratory support, so good laboratory backup was important.

During the whole process oral hygiene and dietary advice were reinforced so that future periodontal and caries problems could be prevented. This periodic review of the patient's oral hygiene and periodontal health must be stressed and maintained in order to achieve long-term success. Psychological health is also an important issue in AI patients¹⁰. Although the appearance of the restorations could have been better, the patient was very happy with the outcome and mastication was satisfactory. Besides achieving the objectives set out at the beginning of the treatment, psychologically she became a happier and more confident person.

References

1. Crawford PJ, Aldred MJ. Anomalies of tooth formation and

- eruption. In: Welbury RW, editor. Paediatric dentistry. Oxford: Oxford University Press; 2005:306-11.
2. Sundell S, Koch G. Hereditary amelogenesis imperfecta. I. Epidemiology and clinical classification in a Swedish child population. *Swed Dent J* 1985;9:157-69.
 3. Wright JT, Robinson C, Shore R. Characterization of the enamel ultrastructure and mineral content in hypoplastic amelogenesis imperfecta. *Oral Surg Oral Med Oral Pathol* 1991;72:594-601.
 4. Aldred MJ, Savarirayan R, Crawford PJ. Amelogenesis imperfecta: a classification and catalogue for the 21st century. *Oral Dis* 2003;9:19-23.
 5. Weinmann JP, Svoboda JF, Woods RW. Hereditary disturbances of enamel formation and calcification. *J Am Dent Assoc* 1945;32:397-418.
 6. Neville BW, Damm DD, Allen CM, Bouquot JE. Oral and maxillofacial pathology. 2nd ed. Philadelphia: Elsevier; 2002:89-94.
 7. Rao S, Witkop CJ Jr. Inherited defects in tooth structure. *Birth Defects Orig Artic Ser* 1971;7:153-84.
 8. Backman B, Holm AK. Amelogenesis imperfecta: prevalence and incidence in a northern Swedish county. *Community Dent Oral Epidemiol* 1986;14:43-7.
 9. Witkop CJ. Hereditary defects in enamel and dentin. *Acta Genet Stat Med* 1957;7:236-9.
 10. Coffield KD, Phillips C, Brady M, Roberts MW, Strauss RP, Wright JT. The psychological impact of developmental dental defects in people with hereditary amelogenesis imperfecta. *J Am Dent Assoc* 2005;136:620-30.
 11. Pindborg JJ. Aetiology of developmental enamel defects not related to fluorosis. *Int Dent J* 1982;32:123-34.
 12. Schulze C. Developmental abnormalities of the teeth and jaws. In: Gorlin RJ, Goldman HM, editors. *Thomas' oral pathology*. 6th ed. St Louis: Mosby; 1970:96-178.
 13. Michaelides M, Bloch-Zupan A, Holder GE, Hunt DM, Moore AT. An autosomal recessive cone-rod dystrophy associated with amelogenesis imperfecta. *J Med Genet* 2004;41:468-73.
 14. Smith BG. Planning and making crowns and bridges. UK: Martin Dunitz; 1998:76-7.
 15. Capp NJ. Occlusion and splint therapy. *Br Dent J* 1999;186:217-22.
 16. Howat AP, Capp NJ, Barrett NV. Colour atlas of occlusion and malocclusion. St. Louis: Mosby Year Book; 1991:181.
 17. Turner KA, Missirlan DM. Restoration of the extremely worn dentition. *J Prosthet Dent* 1984;52:467-74.