Dental management of severe dentinogenesis imperfecta in a mild form of osteogenesis imperfecta

L.X.G. Stephen* / P. Beighton**

Dentinogenesis Imperfecta (DI), in which the teeth are discolored, translucent and brittle, can occur in isolation as a familial trait and as a component of the skeletal dysplasia Osteogenesis Imperfecta (OI). In a Cape Town family, 20 persons in 3 generations had mild OI, with the additional manifestation of severe DI. The family was assessed at the Dental Genetic Unit of the University of the Western Cape and appropriate dental treatment was provided. In this setting, a detailed treatment plan was devised for a severely affected woman. This plan proved to be efficient and cost effective, and the final outcome was pleasing to the patient. Dentinogenesis Imperfecta is not uncommon and may well be encountered in conventional dental practice. The necessary clinical expertise is within the scope of the skills of the general dentist.

J Clin Pediatr Dent 26(2): 131-136, 2002

INTRODUCTION

Osteogenesis Imperfecta (OI) is a heritable disorder of connective tissue in which bone fragility, multiple fractures and sclera blueness are the main manifestations.[1] The teeth of some affected persons are brittle, with a translucent purple or brownish hue. This abnormality, dentinogenesis imperfecta (DI) often necessitates specialised dental management.

More than 400 affected persons have been documented in all populations in Southern Africa[2-4], and it is evident that OI is comparatively common. In order to provide South African colleagues in oral medicine with a perspective of the implications of management of DI in persons with OI, we have documented the dental features of 20 affected individuals in three generations of Cape Town kindred. In this article we summarize the dental features in the family, present a case report which exemplifies the dental complications, and propose a detailed plan for optimal dental management.

PATIENTS AND METHODS

Three decades ago members of a Cape Town family presented with severe dental problems and a mild fracturing tendency. Comprehensive clinical, radiological and genealogical investigations indicated categorisation as a mild form of OI Type I and substantiated an autosomal dominant mode of genetic transmission.[5] The main characteristics were severe DI in the 20 affected individuals (11 males and 9 females in 3 generations), with blue sclerae, a minor fracturing tendency and wormian bones in the cranial sutures. Molecular investigations subsequently revealed linkage with the gene COL1A2, located on chromosome 7q22.1 (vide infra).

In the present study, the family members accepted an offer of a detailed evaluation of their dental changes by LS at the Dental Genetic Unit, Faculty of Dentistry, University of the Western Cape.

DI was very evident in the 20 affected individuals. In these persons, the teeth were all affected, although certain teeth, notably the incisors, showed a greater degree of discoloration. Six persons were treated for their dental condition and of these, 5 reported that DI had been present in all deciduous and permanent teeth. The sixth reported that his deciduous teeth had been normal. Affected teeth had a bluish translucency, with enamel fracturing and severe attrition that is characteristic of DI. Enamel fracturing was particularly prominent in the lower incisors. The occlusal wear was severe, especially on the posterior molars, and resulted in a closed bite as well as reduced facial height in some affected persons.

* Stephen L.X.G., B.Ch.D., Ph.D., Faculty of Dentistry, University of the Western Cape, UWC Dental Special Dental Clinic for Genetic Disorders, Red Cross Children's Hospital, Rondebosch, Cape Town.

** Beighton P., M.D., Ph.D., FRCP., Department of Human Genetics, University of Cape Town, UWC Dental Special Dental Clinic for Genetic Disorders, Red Cross Children's Hospital, Rondebosch, Cape Town

Send all correspondence to Professor LXG Stephen, Faculty of Dentistry, University of the Western Cape, Private Bag X 17, Mitchells Plain, 7785, Cape Town, South Africa.

Tel: +27-21-370-4412
Fax: +27-21-392-3250
Email: LStephen@uwc.ac.za

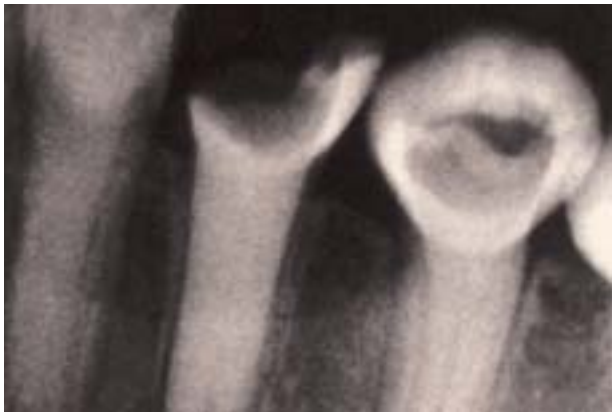

Figure 1. Radiographic features of the typical dentition in this family.

Unlike other forms of OI, the bone fragility in this kindred was a minor feature, which did not overshadow the severe dental anomalies. Affected individuals were thus particularly concerned about their dentition. Nevertheless, many adults were edentulous and it was difficult to motivate younger patients to have restoration and reconstruction of their teeth.

The radiographic features of the teeth were very distinctive. In particular, the crowns were bulbous, the cervical constrictions were marked and the roots were short and blunt. Complete obliteration of the pulp chambers was observed in most teeth and in many there was little or no evidence of root canals (Figure 1). No supernumerary teeth were observed and there was no evidence of root fractures. Dental caries was not particularly prevalent and significant periodontal disease was not diagnosed in any members of the family.

CASE REPORT

Patient CD, a female aged 16 years had the salient features of the condition, notably DI, blue sclerae and wormian bones. She had not fractured any bones and her general health was good.

She reported that her deciduous teeth had been affected, and on examination, the permanent teeth were found to be severely discoloured and brittle. Obvious fractures of the clinical crowns, changes in tooth form as well as marked attrition were prominent features. This resulted in a loss of vertical height (i.e. closed bite) which was accentuated by the patient's prominent mandible, as well as her relative maxillary hypoplasia. This malocclusion caused a "dished in" profile or "reversed occlusion" (Figure 2). There was a reversed overjet and overbite; the mid-third of the face was underdeveloped (Figure 3). Although the anterior crossbite was noted in this individual, other members of the family who were examined did not display similar features nor did the edentulous members of the family report that they had similar malocclusions.

Figure 2. Lateral profile of CD: Pre-treatment. Loss of vertical height of occlusion is evident.

Figure 3. Reversed overjet and reversed overbite.

The frequency of caries was low. Twenty-five teeth were sound and only one tooth required a restoration. Six molar teeth had been extracted on the advice of her dentist. It was difficult to determine whether or not these teeth had been carious, since severe DI may have been confused with dental caries.

Panograph and intra-oral radiographs clearly demonstrated obliteration of the pulp chambers and root canals. There was complete pulpal obliteration in most teeth. The roots were short and blunt and obvious cervical constriction of the crowns was particularly noticeable in the premolar and molar teeth. This factor, coupled with the occlusal wear, contributed to a major alteration in tooth form and structure.

The periodontal status of the patient was acceptable and the severity of the gingivitis was consistent

Treatment Plan. Table I

Phases of the treatment plan	
Phase I	Initial Treatment: - Oral hygiene instruction and motivation Scaling and professional prophylaxis Conservative restoration of tooth No. 26 (amalgam restoration)
Phase II	Correction of vertical height of occlusion and construction of an overdenture for the upper dental arch
Phase III	Porcelain-gold VMK full crowns for the lower dental arch
Phase IV	Recall and maintenance programme

with the amount of plaque and/or calculus present; a diagnosis of moderate gingivitis was recorded. In fact, early mild or moderate gingivitis was noted in the younger members of the family. The adult members of this kindred who were edentulous, reportedly did not have periodontal disease and had extracted their teeth because of the severe discoloration and fracturing. Beside occlusal wear and enamel fractures, no additional factors complicated her oral hygiene. She was very enthusiastic and interested in her oral status; detailed discussions concerning the consequences of tooth loss in a prognathic patient motivated this young female to accept a full reconstruction of her dentition.

Main determinants in the choice of treatment for this patient included: poor aesthetics, marked discoloration and fracturing of the dentition.

The jaw size discrepancy and mid-facial hypoplasia resulted in a crossbite, which made normal functioning (eating in particular) very difficult. The patient was particularly conscious about eating in company since it was often difficult to keep food in her mouth whilst eating. The loss of vertical height of her dentition caused an accentuation of her prominent mandible and resulted in a "dished-in" face.

Objectives of treatment to improve the aesthetic appearance of the dentition: recover the lost vertical dimension of occlusion caused by severe attrition and mid-facial hypoplasia. Prevent further wear of the permanent teeth. Re-establish acceptable facial contours. Improve oral hygiene and home care by providing a dentition, which was easy to maintain.

The above treatment plan was devised after careful determination of vertical height of occlusion. After repeatedly checking the freeway space, it was decided that there was sufficient intermaxillary space available for the construction of an overdenture for the upper arch, without occlusal reduction or preparation of copings on the supporting teeth.

Phase I. Initial treatment

The initial routine dental visits included, history taking and completion of records. Time was allocated for oral hygiene instruction and motivation as well as scaling and polishing. Oral hygiene status is of prime importance especially in a patient with extensive crowns and overdenture prosthesis. During this phase of treatment, tooth 26 was restored, and all sharp edges of other teeth were smoothed. When it was apparently that meticulous oral hygiene was being done, the expectations of the patient were analysed and various treatment options were discussed. A detailed treatment plan was presented, with due consideration given to the time and co-operation that was expected if an acceptable outcome was envisaged.

Phase II. Construction of maxillary overdenture

The decision to construct a maxillary overdenture was based on the following factors:

1. The vertical height of occlusion could be corrected by this method with little damage to the remaining teeth. (No other conventional treatment such as crown preparation of supporting teeth was indicated).
2. The grossly malaligned "reverse bite" or "cross-bite" could be corrected. Some of the additional bulk of the overdenture compensated for the maxillary hypoplasia (Figure 4), and this resulted in an aesthetically pleasing result.
3. To achieve ease of placement of the overdenture, minimal preparation of the upper teeth (without the use of local anaesthesia) was necessary. This process basically involved reducing undercuts of the very bulbous supporting teeth. Sharp ridges of fractured teeth were smoothed.
4. The procedure would be completely reversible since no copings were constructed and extensive preparation of the natural dentition was not necessary.
5. The cost of constructing an overdenture is economical if compared to the complete rehabilitation of the dentition using gold and porcelain VMK crowns.

Step 1: Prior to construction of the overdenture, an occlusal bite plate was constructed in order to determine the patient's tolerance of a decrease in freeway space. This approach also enabled the clinician to determine the optimal vertical height of occlusion. Adequate function, muscular tolerance and aesthetics were important clinical factors that were also considered in determining the correct vertical height. Initially, an acrylic bite plate was constructed, merely to disocclude the dentition; subsequently, cold cure clear acrylic was added at weekly intervals until the optimal height of occlusion was attained. This was a cost-effective method of determining the correct vertical height and clinical appointments were short.

Step 2: Once the correct height of occlusion had been ascertained, a face-bow was used to transfer the measurements and set a semi-adjustable Hanau articulator. These factors were used to construct an overdenture.

Step 3: An upper overdenture was constructed using standard dental procedures. Impressions were taken using a polyether rubber impression material. A "try-in" stage produced pleasing results and the denture was completed in three visits.

At the final visit, a thorough prophylaxis was performed, all sharp edges of teeth were reduced and smoothed and a fluoride treatment (1.23% acidulated phosphate fluoride gel) was applied. The patient was given detailed instruction on denture care.

Following a few minor adjustments, the patient accepted the aesthetics and was comfortable wearing the upper denture. There was an overall improvement in function and aesthetics, and the profile of the patient was greatly improved.

Figure 4. Completed overdenture. The aesthetics of maxillary dentition are masked and the vertical height of occlusion is corrected.

Phase III. Reconstruction of lower arch using porcelain-VMK full crowns

Technically, this was the most challenging phase of therapy in this dentition.

A decision was made to use full crowns (VMK porcelain veneered onto gold copings) for the following reasons:

1. The correct height of occlusion had been determined, and further attrition was undesirable. Full VMK crowns would prevent this loss of vertical dimension.
2. The aesthetic results would be pleasing, since the discoloration would be entirely masked.
3. The interdental spaces could be closed by means of increasing the crown width.

Following the construction, placement and adjustment of an overdenture for the maxillary arch, and prior to commencing the treatment of the lower dental arch, an interim period enabled further evaluation of patient co-operation. This approach was imperative for evaluation of oral hygiene compliance prior to construction of the expensive lower crowns.

The construction of single crowns was undertaken using standard dental procedures. Following initial impressions, a diagnostic wax-up was provided so as to gain the patient's opinion and approval. Thereafter, the crowns were prepared using fine-cutting diamond burs. It must be emphasised that **no local anaesthesia was necessary** and the patient was fully comfortable during the preparations of the crowns.

Due to the alterations in crown form, it was decided that 90 degree butt joints would be used on the anterior teeth (Figure 5). This approach enabled the use of porcelain margins at the cervical regions of the preparations. Excessive reduction of tissue at an already weakened region of the tooth was avoided. Since the margins of the premolar crowns were not visible during normal function, and due to the marked cervical construction of these teeth, a chamfered preparation was preferable, allowing metal margins to be placed on these teeth. Thus, minimal preparation of these teeth was necessary. All margins were placed supragingivally, which prevented further compromise of the oral hygiene and resulted in pleasing aesthetics (Figure 6).

Temporary crowns were used between visits and within 14 days the permanent crowns were placed. Initially, temporary cement was used to lute the crowns. This allowed a period for re-evaluation and assessment of any incidental problems. Subsequently, the patient was recalled and after 3 months the crowns were permanently cemented.

Phase IV. Recall and maintenance

Recall visits were scheduled every 3 months. At these visits, the prostheses, the supporting teeth as well as the gingival and periodontal tissues were evaluated. Special attention was paid to the crown margins and the entire dentition was treated with 1.23% APF fluoride

Figure 5. Crown preparations of lower anterior teeth; premolar crowns are in situ and the maxillary overdenture is placed.

Figure 6. Permanent restoration of the dentition.

gel. After six months, the occlusion and oral hygiene status had stabilised and no further adjustments to the dentures or crowns were needed.

The correction of facial height was well tolerated and the patient reported efficient mastication and did not experience any TMJ pain or dysfunction. In fact, this individual has subsequently followed up for several years and has been functional and comfortable.

Following the successful restoration and reconstruction of the dentition, her siblings also presented for treatment. Using standard dental procedures, porcelain jacket crowns were constructed, which resulted in an acceptable aesthetic appearance (Figure 7).

DISCUSSION

The nosology of DI and OI is complex, but in essence the combination of DI and an autosomal dominant form of mild to moderate OI with sclera blueness is categorised as a sub-group of OI Type I.[6,7] It would be appropriate to regard the kindred documented in this article as having this form of OI.

OI type I is the result of defects in the alpha I or alpha II chains of the collagen molecule, which influence the production and/or assembly of type I collagen. In turn, these abnormalities are the consequence of mutations in the corresponding genes, COL1A1 and COL1A2. The former had been mapped to chromosome 17q21.31-q22 while the latter is situated at 7q22.1. The family documented in this article had been shown to have a mutation in COL1A2, in keeping with the diagnosis of OI.

It is relevant that DI can occur as an isolated trait, in persons who are otherwise clinically normal. Witkop and Rao[8] suggested that isolated familial DI should be termed "opalescent dentine". In an alternative nosology, Shields *et al*.[9] classified DI in OI as "DI type I", with isolated DI as "DI type II" (MIM 125490) and "DI type III" (MIM 125500), on a basis of differing severity in large kindred documented in the USA. DI-II became known as the "Shields type" while DI-III was termed the "Brandywine type", named after the region in Maryland, USA, where an affected kindred originated. Subsequently, Witkop[10] commented that the same family name occurred in both kindred, and suggested that DI-II and DI-III were the same entity. Linkage studies have indicated that the genes for DI-II and III are both on the long arm of chromosome 4 (4q21-q25), and that the two putative conditions are either the same condition or result from different mutations within the same gene.[11-13] In any event, isolated DI is a different entity from DI in OI, with different causative genetic mechanisms.

Although the major manifestations of OI are the result of abnormalities of collagen in the skeleton, sclera and dentine, other connective tissues may also be involved.[14] In this way, fragility of the gingiva might predispose to periodontal disease. The mild gingivitis present in the South African patients could be a reflection of this process.

Various treatment modalities have been discussed in the literature.[15-17] These included extraction of teeth and placement of full dentures, porcelain veneers, composite veneers, and full crowns, as well as combinations of these prosthetic constructions. The treatment plan embarked upon for the South African patient had many advantages. It was cost effective in terms of the laboratory and clinical time as well as the materials that were used. Patient comfort was optimal and local anaesthesia was unnecessary. There is a marked decrease in the transmission of nervous sensation due to the obliteration of pulpal tissue and the presence of poorly defined, tortuous occluded dentinal tubules. In fact, it is seldom necessary to anaesthetise these patients for dental restorative procedures. The clinical expertise needed for this treatment is within the scope of a general dentist and it was not necessary to refer the

Figure 7. Reconstruction of dentition resulted in a pleasing aesthetic appearance.

patient for any specialised prosthodontic procedures. The fact that the oral hygiene status was acceptable, (after a few visits for instruction and prophylaxis) coupled with the low caries rate, proved to be very advantageous. As highlighted earlier, each decision made in drawing up a treatment plan for this young, self-conscious patient was well founded and justifiable. All the objectives set out initially were achieved and the patient was very pleased with the permanent cosmetic result.

A few pertinent facts regarding the treatment of this patient are relevant. Firstly, the patient was young, intelligent, self-conscious, co-operative and available for this extensive treatment plan. Given these factors and the demand for highly complicated aesthetic dentistry, there was little option but to offer her the best treatment without compromise.

REFERENCES

1. Tsipouras P. in Beighton P (ed) McKusick's Heritable Disorders of Connective Tissue. 5th ed. St Louis: Mosby, p. 218-314, 1993.
2. Beighton P, Spranger J, Versfeld G. Skeletal complications in osteogenesis imperfecta, a review of 153 South African patients. S Afr Med J 64: 565-568, 1983.
3. Beighton P, Versfeld GA. On the paradoxically high relative prevalence of osteogenesis imperfecta type III in the black population of South Africa. Clin Genet 27: 398-401, 1985.
4. Viljoen D, Beighton P. Osteogenesis imperfecta type III: an ancient mutation in Africa? Am J Med Genet 27: 907-912, 1987.
5. Beighton P. Familial dentinogenesis imperfecta, blue sclerae, and wormian bones without fractures: another type of osteogenesis imperfecta? J Med Genet 18: 124-128, 1981.
6. Levin LS, Brady JM, Melnick M. Scanning electron microscopy of teeth in dominant osteogenesis imperfecta. Am J Med Genet 5: 189-199, 1980.
7. Paterson CR, McAllion S, Miller R. Heterogeneity of osteogenesis imperfecta type I. J Med Genet 20: 203-205, 1983.
8. Witkop CJ Jr., Rao SR. Inherited defects in tooth structure. Birth Defects Orig Art Ser 7: 153-184, 1971.
9. Shields ED, Bixler D, El-Kafrawy AM. A proposed classification for heritable human dentine defect with a description of a new entity. Arch Oral Biol 18: 543-553, 1973.
10. Witkop CJ Jr. Hereditary defects of dentin. Dent Clin N Am 19: 25-45, 1975.
11. Broughman JA, Halloran SL, Roulston D et al. An autosomal dominant form of juvenile periodontitis: its localization to chromosome 4 and linkage to dentinogenesis imperfecta and Gc. J Craniofac Genet Dev Biol 6: 341-350, 1986.
12. Dean JA, Hartsfield JK Jr., Wright JT, Hart TC. Dentin dysplasia, type II linkage to chromosome 4q. J Craniofac Genet Dev Biol 17: 172-177, 1997.
13. MacDougall M, Jeffords LG, Gu TT et al. Genetic linkage of the dentinogenesis imperfecta type III locus to chromosome 4q. J Dent Res 78: 1277-1282, 1999.
14. Raff ML, Craigen WJ, Smith LT, Keene DR, Byers PH. Partial COL1A2 gene duplication produces features of osteogenesis imperfecta and Ehlers-Danlos syndrome type VII. Hum Genet 106: 19-28, 2000.
15. Giansanti JS, Allen JD. Dentin dysplasia, type II, or dentin dysplasia, coronal type. Oral Surg 38: 911-917, 1974.
16. Wald C, Diner H. Dysplasia of the dental pulp: report of a case. J Dent Child 41: 212-215, 1974.
17. Nayer AK, Latta JB, Soni NN. Treatment of dentinogenesis imperfecta in a child: report of a case. J Dent Child 48: 453-455, 1981.

ACKNOWLEDGEMENTS

The laboratory procedures and manufacturing of the porcelain veneers were undertaken by Deon Valentine of the University of the Western Cape. We are grateful for his contribution and to Yolanda Erasmus and Greta Beighton for typing the manuscript.