

Dentinogenesis Imperfecta Associated with Osteogenesis Imperfecta: Report of Two Cases

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Osteogenesis imperfecta (OI) is a heritable systemic disorder of the connective tissue. Dentinogenesis imperfecta (DI), which is sometimes an accompanying symptom of OI, belongs to a group of genetically conditioned dentin dysplasias and is characterized clinically by an opalescent amber appearance of the dentin. Although the teeth of DI cases wear more easily and excessively compared to normal teeth, they do not appear to be more susceptible to dental caries than normal teeth. Two cases of DI associated with OI are presented in this paper, with 1 case suffering from nursing bottle caries. The purposes of this paper are to present the dental and skeletal characteristics of moderately and mildly involved DI associated with OI, and to discuss the possible methods of dental treatment. Patients with OI and opalescent teeth should be evaluated as soon as the deciduous teeth erupt; immediate dental involvement and oral hygiene instruction can be of help in reducing the necessity of extensive dental care. (*Chang Gung Med J* 2003;26:138-43)

Key words: dentinogenesis imperfecta, osteogenesis imperfecta, nursing bottle caries.

Osteogenesis imperfecta (OI) is a heterogeneous group of genetic disorders that affect connective tissue integrity. Most forms of OI are the result of mutations in the genes (COL1A1 and COL1A2) that encode the pro alpha 1 and pro alpha 2 polypeptide chains of type I collagen.⁽¹⁾ Tissues in which the principal matrix protein is type I collagen (mainly bone, dentin, sclerae, and ligaments) can be affected. The resultant abnormalities include blue sclera, rigidity of the osseous tissue, hearing loss, dentinogenesis imperfecta (DI), growth deficiency, laxity of the joints, and any combination of these characteristics.⁽²⁾ The incidence of OI in infancy is about 1 per 20,000-30,000 in an Australian study.⁽³⁾

There are extreme phenotypic variations within the OI population. Four types of OI including mild, perinatal lethal, progressive deforming, and moderately severe were classified according to clinical,

genetic, and radiographic criteria.⁽⁴⁾ Each of the 4 types of OI is further subdivided on the basis of the absence or presence of DI.⁽⁵⁾ However, many OI patients still cannot be easily assigned to any 1 of the 4 classes because of the broad spectrum and complexity of molecular abnormalities resulting in OI.⁽¹⁾

Molecular genetic studies of OI have identified more than 150 mutations of the COL1A1 (17q21.3-q22) and COL1A2 (7q22.1) genes. Various systemic treatments for OI (gene therapy or cell therapy) have been attempted, but these interventions were either inconclusive or are still experimental.⁽⁶⁾

Dentinogenesis imperfecta (DI) is characterized clinically by opalescent and translucent dentin due to a mesenchymal defect. The primary teeth are more severely affected than is the permanent dentition. The color of the teeth varies from opalescent gray or brown to yellow, and both upper and lower denti-

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tions are involved.⁽⁷⁾ Radiographically, the crowns of the teeth are bulbous with marked cervical constrictions, and pulp chambers become obliterated over a period of time.^(8,9) The thickness and radiodensity of the enamel are normal. Significant attrition can be seen over a short period of time.⁽¹⁰⁾ The reported incidence in the US is 1 in 8000 births.⁽¹¹⁾

There are 3 types of dentinogenesis imperfecta. Type I is associated with osteogenesis imperfecta. Type II is the so-called classical heredity opalescent dentin. Type III is the Brandywine form, named for the city of Brandywine, MD, where there was a large population of patients with this disorder. Type III tends to be less severe than type II.

Dentinogenesis imperfecta has an autosomal dominant pattern of inheritance. Ball et al. found that the type II locus is located on 4q13.⁽¹²⁾ Types II and III are different expressions of the same gene.⁽¹³⁾ The genes for various forms of OI have been localized to 17q21.31-q22.05 and 7q21.3-9 22.1, and DI type I may be associated with these genes.

Some patients with OI display no abnormalities in the dentition, whereas others manifest significant dentinal involvement. Other oral manifestations associated with OI include midfacial hypoplasia, a shortened maxilla with normal mandibular length, class III malocclusion, and posterior crossbite.^(14,15) There are no valid statistics about DI-associated OI in Taiwan.

The purposes of this article are to present the dental and craniofacial characteristics of these 2 cases of moderately and mildly involved OI associated with DI and to discuss the dental management.

CASE REPORTS

Case 1

A 5-year-old Taiwanese girl was referred by her pediatrician to the pediatric dental clinic at Chang Gung Memorial Hospital, Kaohsiung; she had complained of a toothache for several days. She presented facial symmetry with a concave profile and a class III skeletal pattern.

Her medical history revealed that she had had enlarged posterior fontanelles, a dilated ventricle, and a fracture of the right femoral shaft at birth. A diagnosis of OI was made. Although the child was mentally normal, her physical development was abnormal. Physical examinations showed that she

had limited motility, bowing of the long bones, shortness of the neck, kyphoscoliosis, and protuberance of the sternum.

An intraoral examination revealed multiple dental decay and attrition of the primary teeth. The rampant caries found in the deciduous teeth was possibly a result of a nighttime nursing bottle habit. Her remaining intact teeth showed an opalescent translucent hue.

A panoramic radiograph showed that the intact teeth had bulbous crowns and small pulp chambers lacking pulp horns when compared to normal teeth. Those teeth with severe destruction presented a "shell" appearance of enamel and dentin surrounding a large pulp chamber and root canals. Three permanent premolars (15, 35, and 44) were congenitally missing (Fig. 1). A cephalometric radiograph revealed a shortened maxilla and midfacial hypoplasia.

The patient was diagnosed as OI type IV associated with DI. For dental treatment, we had to put her under general anesthesia because of her uncooperative behavior. The dental treatment procedures included: (a) stainless steel crown on 85; (b) composite resin restoration on the occlusal surface of 36; (c) extraction of severely destructed 55, 54, 53, 52, 51, 61, 62, 63, 64, 65, 74, 75, and 84; and (d) 1.23% acidulated phosphate fluoride application.

At the subsequent 1-year oral examination, the patient showed a typical Class III malocclusion. The upper central incisors had erupted and showed an opalescent brown tooth color (Fig. 2).



Fig. 1 Panoramic radiography of case 1. Note that the teeth with severe destruction present a "shell" appearance.



Fig. 2 Opalescent tooth appearance with Class III malocclusion on the subsequent 1-year oral examination in case 1.



Fig. 3 Case 2. Both the deciduous and permanent dentitions reveal an opalescent brown to gray color. The deciduous dentition is abraded.

Case 2

This 7.5-year-old Taiwanese girl was brought by her father to the pediatric dental clinic complaining of the ugly appearance of her teeth. She also had a fractured right leg which was fixed in a plaster cast. Her medical history revealed that she had had a left clavicle fracture and bruising over the elbow at birth. She had experienced sustained frequent tibia fibular fractures since age of 6.

An extraoral examination revealed a symmetrical and straight profile with a Class I skeletal pattern. Blue sclera was also noted. An intraoral examination revealed a brown to gray opalescent tooth color in both the deciduous and permanent dentitions; we learned that her father and the father's uncle had been similarly affected. The crowns of the primary teeth were short and abraded. The incisors were edge to edge and the bilateral buccal segments were in a crossbite (Fig. 3).

A panoramic radiograph showed that the pulp chamber and root canals of the deciduous teeth had been partially or completely obliterated. The permanent teeth lacked pulp horns. The junctions of the crowns and roots were more constricted than those of normal teeth (Fig. 4).

Dentinogenesis imperfecta involving both the deciduous and permanent dentition was diagnosed in this case. She was referred to the pediatric heredity and endocrine department for a general evaluation, and a diagnosis of type I osteogenesis imperfecta (Sillence classification) was confirmed.⁽⁴⁾

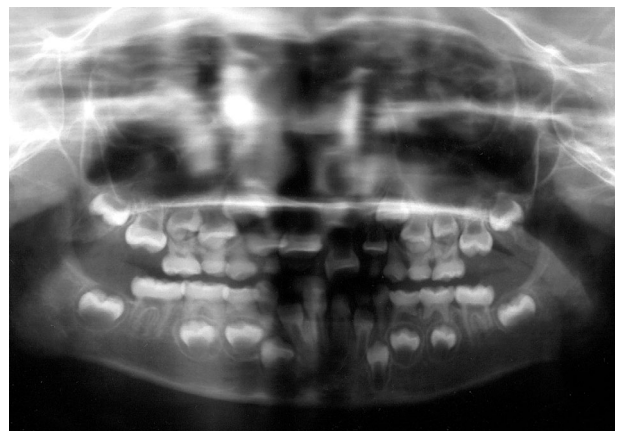


Fig. 4 Panoramic radiography of case 2. Note that the pulp chamber and root canals have been partially or completely obliterated.

Due to only slightly abraded teeth seen in case 2, the dental treatment plan was to observe her at 6-month intervals. Placement of artificial crowns will be considered if these teeth are seen to be rapidly breaking down or fracturing.

DISCUSSION

The aims of dental treatment for children with DI associated with OI are to ensure favorable conditions for eruption of the permanent teeth and normal growth of the facial bones and temporomandibular

joints.⁽¹⁶⁾ The oral problems in the first case included rampant caries with multiple residual roots, collapse of occlusal height, and hypodontia. A "shell" appearance of the deciduous teeth (74,75,84) was also found. Levin (1981) described how the pulp chambers and root canals in DI may be patent on eruption, and even be larger than those of normal teeth.⁽¹⁷⁾ Severe dental caries does not seem to be a major problem in patients with DI and OI. However, caries appears to inhibit the obliteration process of the pulp chamber, which results in a wider tooth canal space than with a typical obliterated canal space in DI. Children diagnosed as having OI should be seen by a dentist as soon as possible after the eruption of the deciduous anterior teeth in order to determine whether there is DI involvement. Dental care for DI includes caries prevention, attrition observation, and monitoring of skeletal development. If the deciduous teeth begin to wear, placement of artificial crowns is recommended before excessive loss of tooth structure occurs.⁽¹⁷⁾

In case 1, almost all of the primary teeth were severely decayed and were removed under general anesthesia. Such rampant caries was mostly due to an inappropriate nursing bottle habit. An overlay denture was indicated for rehabilitating the oral function and occlusion in case 1. However, it was not put in place because of her uncooperative behavior.

In case 2, the exposed dentin of 52, 53, 62, and 63 was seen to wearing toward the gingival line, and the primary molars exhibited excessive attrition with enamel fracture. However, we did not plan on crowning these primary molars to rehabilitate the occlusal height, because the caries rate was not high compared to case 1, and the permanent first molars were intact. Schwartz and Tsipouras concluded⁽¹⁵⁾ that dental attrition in DI was less severe in the permanent than in the primary dentition. Unusual wear does not occur as frequently and to such a great extent in permanent as in deciduous teeth. Further esthetic considerations for improving tooth color may be full-jacket crowns or porcelain veneer crowns on the anterior teeth.⁽⁵⁾ Although bonding of the resin to the defective teeth structure is theoretically compromised, it is clinically successful in most patients.⁽⁵⁾

Jensen et al. found that the posture, weight, and size of the head were abnormal in the OI population, which might contribute to the development of maloc-

clusion. The more-severe abnormalities of craniofacial features were associated with the more-severe types of OI in adult patients.⁽¹⁸⁾ O'Connell et al. reported that class III dental malocclusion occurred in 70% to 80% of types III and IV of the OI population, with a high incidence of anterior and posterior crossbites and open bites.⁽¹⁰⁾ This was true for case 1 who had a class III skeletal pattern and a retrusive tendency in the maxilla. Orthodontic and surgical procedures for correcting the malocclusion are very difficult because of the easy fracturing tendency in OI patients. Maintenance of the deciduous teeth is therefore particularly important to insure normal alignment of the permanent teeth and to reduce the necessity of extensive orthodontic care. Case 2 had a class I skeletal pattern, but the mesiodistal dimension of the primary molars had decreased due to excessive attrition. Potential crowding of the buccal segments was predicted due to the decreased arch length. Orthodontic treatment in patients who suffer from DI associated with OI has not been reported until now. However, orthodontic treatment has been successfully performed in patients with different degrees of DI.^(19,20)

In conclusion, OI consists of heritable systemic disorders of the connective tissue. DI is a possible accompanying symptom of OI and belongs to the group of genetically conditioned dentin dysplasias. Patients with OI and opalescent teeth should be evaluated as soon as the deciduous teeth erupt, so that an attempt can be made to prevent loss of tooth structure.

REFERENCES

1. Niyibizi C, Smith P, Mi Z, Robbins P, Evans C. Potential of gene therapy for treating osteogenesis imperfecta. *Clin Orthop Relat res* (379 suppl) 2000;S126-33.
2. Marini JC. Osteogenesis imperfecta: comprehensive management. *Adv Pediatr* 1988;35:391-426.
3. Sillence DO. Osteogenesis imperfecta: an expanding panorama of variants. *Clin Orthop Rel Res* 1981;159:11-25.
4. Sillence DO, Senn A, Danks DM. Genetic heterogeneity in osteogenesis imperfecta. *Am J Med Genet* 1979;16:101-16.
5. Sillence DO. Osteogenesis imperfecta nosology and genetics. *Ann NY Acad Sci* 1988;543:1-15.
6. Kocher MS, Shapiro F. Osteogenesis imperfecta. *J Am Acad Orthop Surg* 1988;6(4):225-36.
7. Shafer WG, Hine MK, Levy BM. Developmental distur-

- bances of oral and paraoral structures. In: A Textbook of Oral Pathology. 4th ed. Philadelphia: W.B. Saunders Co., 1983:58-61.
8. Pindborg JJ. Dental aspects of osteogenesis imperfecta. *Acta Pathol Microbiol Scand* 1947;24:47-58.
 9. Stafne EC. Dental roentgenologic manifestations of systemic disease. II. Developmental disturbances. *Radiology* 1952;58:507-16.
 10. O'Connell AC and Marini JC. Evaluation of oral problems in an osteogenesis imperfecta population. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 1999;87:189-96.
 11. Witkop CJ. Genetics and Dentistry. *Eugen Quart* 1958; 5:15-21.
 12. Ball SP, Cook PJJ, Mars M, Buckton KE. Linkage between dentinogenesis imperfecta and Gc. *Ann Hum Genet* 1982;46:35.
 13. MacDougall M, Jeffords LG, Gu TT, Knight CB, Frei G, Reus BE, Otterud B, Leppert M, Leach RJ. Genetic linkage of the dentinogenesis imperfecta type III locus to chromosome 4q. *J Dent Res* 1999;78(6):277-82.
 14. Isshiki Y. Morphological studies on osteogenesis imperfecta, especially in teeth, dental arch and facial cranium. *Bull Tokyo Dent Coll* 1996;7:31-49.
 15. Schwartz S, Tsipouras P. Oral findings in osteogenesis imperfecta. *Oral Surg Oral Med Oral Pathol* 1984;57: 161-7.
 16. Ranta H., Lukinmaa PL, Waltimo J. Heritable Dentin Defects: Nosology, pathology, and treatment. *Am J Med Genetics* 1993;45:193-200.
 17. Levin LS. The dentition in the osteogenesis imperfecta syndromes. *Clin Orthop Related Res* 1981;159:64-74.
 18. Michael DC. Dentinogenesis imperfecta: a case report. *Am J Orthod Dentofacial Orthop* 1998;113:367-71.
 19. Battagel JM. Dentinogenesis imperfecta: an interdisciplinary approach. *Br Dent J* 1988;165:329-31.
 20. Jensen BL, Lund AM. Osteogenesis imperfecta: clinical, cephalometric, and biochemical investigations of OI types I, III, and IV. *J Craniofac Genet Dev Biol* 1997;17:121-32.

牙本質發生不全合併成骨不全：二例報告

蔡佳玲 林瑩澤 林雅婷

成骨不全是遺傳性中胚葉的缺陷，而牙本質發生不全是生牙器 (odontogenic apparatus) 的中胚層部分發生障礙，兩者可能聯合出現。牙本質發生不全的牙齒外觀呈現特殊的透明性及乳光色澤，顏色從灰白色到黃褐色都有，比正常牙齒更容易磨損，但這類牙齒對齲齒的感受性並不比正常者大，反而較低。我們報告兩個年紀在早期混合齒列期的牙本質發生不全聯合成骨不全病例，其中一位患有奶瓶性齲齒。報告目的在於提出中等到輕微程度的成骨不全同時患有牙本質發生不全患者的口腔及顱骨特徵，並討論可能的牙科治療。成骨不全患者在乳牙剛萌出時即應由牙醫師做評估是否同時患有牙本質發生不全，牙科即時的介入衛教及處理將有助於減少廣泛而複雜的牙科治療。(長庚醫誌 2003;26:138-43)

關鍵字：牙本質發生不全，成骨不全，奶瓶性齲齒。

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