

# Dentinogenesis Imperfecta: A Case Report of Five Patients in the Same Family Group

Dentinogenesis Imperfecta: Un Reporte de Caso de  
Cinco Pacientes en el Mismo Grupo Familiar

Gonzalo Muñoz<sup>\*</sup>; Fernanda Ojeda<sup>\*\*</sup>; Víctor Sáez<sup>\*\*\*</sup>; Eduardo Borie<sup>\*\*\*\*</sup> & Evelyn Borie-Echevarría<sup>\*\*\*\*\*</sup>

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**ABSTRACT:** Dentinogenesis imperfecta (DI) is a type of dentin dysplasia that affects the dentin structure of one or both dentitions, which may be classified in three types. The aim of this report was to show the clinical and radiographic features of the four cases of DI in the same family group. Five brothers were checked clinically and radiographically. Two individuals were diagnosed, by their phenotypic features and medical history, with DI type I; two of them with DI type II and one case without signs of DI. It is important to know the features of dentinogenesis imperfecta to perform a comprehensive dental care, including the right diagnosis and an effective treatment plan.

**KEY WORDS:** dentinogenesis imperfecta, tooth alteration, family, dental development disturbances.

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## INTRODUCTION

Tooth development results in several interactions between the oral epithelium and underlying mesenchymatic tissue during odontogenesis (Borie *et al.*, 2010). During the whole process, changes that interfere with normal tooth formation may happen, disrupting the histodifferentiation, apposition, and tissue calcification (Abukabbos & Al-Sineedi, 2013). The etiology factors involved in dental development disturbances can be hereditary or genetic-environmental (Martín-Gonzalez *et al.*, 2012), altering the enamel (amelogenesis), dentin (dentinogenesis), or all dental tissues from the second month of intrauterine life. The type of development abnormality depends on the stage of tissue development (Martín-Gonzalez *et al.*).

The genetic factor is related to the mutation of one or more genes involved in the amelogenesis or dentinogenesis process, which cause anomalies and

dysplasias. The genes involved in amelogenesis (ENAM, MMP20, and KLK4) and dentinogenesis (DSPP) are highly specific for teeth, so their mutations produce non-syndromic tooth alterations of non-syndromic tooth development, that is, without observing anomalies in other structures (Martín-Gonzalez *et al.*; de La Dure-Molla *et al.*, 2015).

Dentinogenesis imperfecta is a type of dentin dysplasia, being an autosomal dominant hereditary disorder that affects the dentin structure of one or both dentitions. It is originated in the histodifferentiation during odontogenesis process, being a localized form of mesodermal dysplasia characterized by an alteration of the dental proteins (Surendra *et al.*, 2013; Devaraju *et al.*, 2014). DI types II and III are caused by a defect in dentin sialophosphoprotein gene (DSPP) (Kim & Simmer, 2007).

<sup>\*</sup> Student of Faculty of Dentistry, Universidad de La Frontera, Chile.

<sup>\*\*</sup> Private Practice, Panguipulli, Chile.

<sup>\*\*\*</sup> Centro Salud Familiar (CESFAM), Hualpin, Chile.

<sup>\*\*\*\*</sup> CICO Research Centre, Dental School, Universidad de La Frontera, Temuco, Chile.

<sup>\*\*\*\*\*</sup> Escuela de Odontología, Cátedra Odontología Integral del Niño I y II, Universidad Mayor sede Temuco, Temuco, Chile.

The DI is characterized as an opalescent dentin due to the translucent appearance in the affected teeth (Min *et al.*, 2014). Its diagnosis is primarily based on clinical and radiological findings and according to these, DI is classified into 3 types described by Shields *et al.* (1973): Type 1 is associated with osteogenesis imperfecta (OI); Type 2, is not associated with OI, presents similar clinical and radiological features; and Type 3 is also called Brandywine (Trejos *et al.*, 2007; Rabassa *et al.*, 2011).

The purpose of this report was to show the clinical and radiographic features of the four cases of dentinogenesis imperfecta within the same family group.

## CASES REPORT

A clinical study was carried out with five patients between 4-12 years old, treated in the Pediatric Clinics of Universidad Mayor (Temuco, Chile) and diagnosed with dentinogenesis imperfecta (DI). The informed consent was signed by the parents of the patients. In this case report, patients of the same family group whose father, the paternal grandfather, and the paternal aunt diagnosed with this same condition were included. The final diagnosis was based on the phenotypic features and medical diagnosis performed through clinical diagnosis, photographs, patient's medical history and panoramic radiographs. The cases of five brothers are summarized in Table I and are explained in detail below:

**Case 1:** The first case was a 4 year-old female patient with no history of fractures. A round face and concave facial profile was identified during the facial examination. The primary dentition completed with structural alterations and yellow-brown color marks was identified during the intraoral examination (Fig. 1A). At a frontal position, attrition, anterior open-bite, poste-

rior cross-bite at the right side, and dental midline deviated 1.5 mm to the right side were observed. At the sagittal plane, the individual exhibited an open-bite with an overbite of -4 mm and overjet of -2 mm. The radiographic evaluation showed the primary dentition completed and permanent dentition with pathognomonic signs of DI (Fig. 1A). The final diagnosis was DI type II.

**Case 2:** The second case is a male patient, 6 years old, with a history of fractures, two in the forearm, two in the femur, and one in the clavicle. A round face, concave facial profile and gray sclera were identified during the facial examination. During the intraoral examination, mixed dentition with structural alterations and yellow-brown color marks were exhibited (Fig. 1B). At a frontal plane, attrition, anterior cross-bite between the teeth (62 and 72), and dental midline deviation of 2 mm to the right side were identified. At the sagittal plane, an anterior cross-bite with an overbite of -2 mm and overjet of 0 mm was observed. The radiographic evaluation showed the permanent dentition with pathognomonic signs of DI (Fig. 1B). Dental agenesis of teeth 14, 24, 25, 35 and 45 was identified. Development asynchrony of teeth 11 and 22 was observed with a horizontal mesiodens at 21-tooth region. The final diagnosis was DI type I.

**Case 3:** An 8-year-old male patient without history of fractures came to the clinic. Facial examination showed a long face and convex facial profile. Mixed dentition without structural alterations and normal color during intraoral examination were revealed (Fig. 2A). At frontal vision, an anterior open-bite, posterior cross-bite at the right side, and dental midline with 4mm deviation to the right side were observed. At sagittal plane it was identified an overbite of -2 mm and overjet of -0 mm. The radiographic evaluation observed a mixed dentition (Fig. 2A). The permanent dentition showed normal development, gemination tooth of 31, and lack of teeth space for eruption of 13 and 23. The final diagnosis was not compatible with any type of DI.

Table I. Features and diagnosis of each case in the same family group.

Case	Sex	Age	Tooth alterations	Radiographic pathognomonic signs	History of fractures	DI Diagnosis*	DI type
1	F	4	Yes	Yes	No	Yes	Type II
2	H	6	Yes	Yes	Yes	Yes	Type I
3	H	8	Yes	Yes	No	No	No
4	H	11	No	No	No	Yes	Type II
5	F	12	Yes	Yes	Yes	Yes	Type I

\* Diagnosis based on fenotypic features.



Fig. 1. Clinical and radiographical features of case 1 (A), 2 (B) and 4 (C).

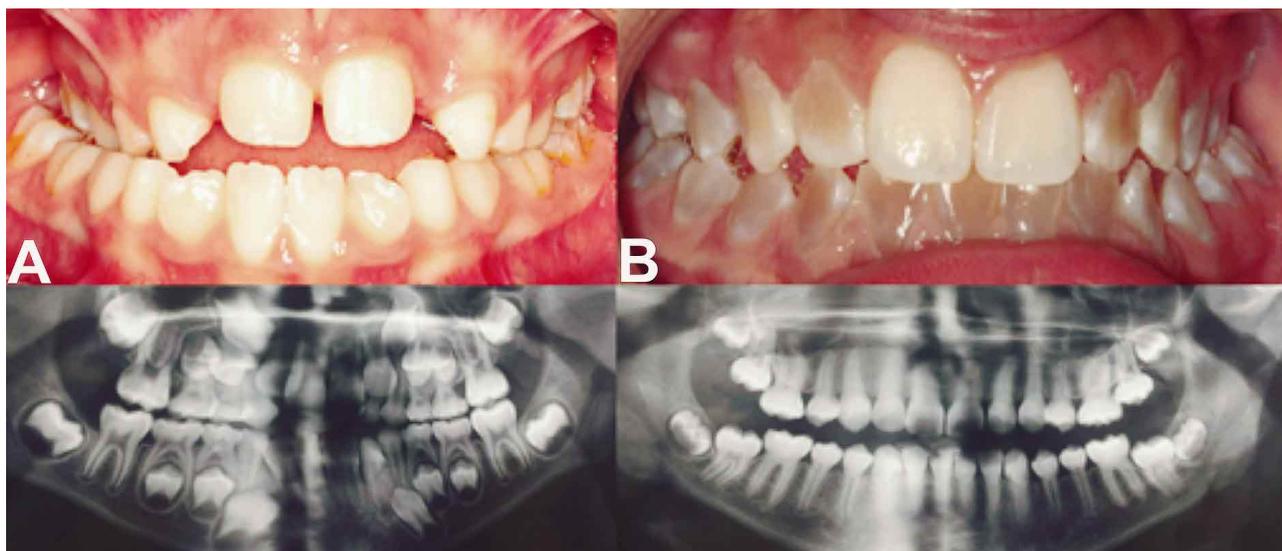


Fig. 2. Clinical and radiographical features of case 3 (A), and 5 (B).

**Case 4:** The next case is a male patient, 11 years old, without history of fractures. The patient showed a round face and convex facial profile during the facial examination. The intraoral examination revealed a permanent dentition with yellow-brown color (Fig. 1C). The frontal vision identified an anterior dental midline that deviated 2.5 mm to the right side. At the sagittal plane, a class I Angle molar was identified at the right side while the left side presented a class III. The radiographical evaluation evidenced a permanent dentition with pathognomonic signs of DI (Fig. 1C). The final diagnosis was DI type II.

**Case 5:** The last case was a 12 year-old female patient with a history of fractures, one in the forearm and two at the femur. The patient exhibited round face, convex facial profile and gray sclera during the facial examination. The intraoral examination evidenced mixed dentition with yellow-brown color, except on teeth 11 and 21 (Fig. 2B). At the frontal plane, she showed

left cross-bite and dental midline that deviated 5 mm to the right side. At the sagittal plane, a class I Angle molar was identified at the right side while the left side presented a class II. The radiographical evaluation evidenced the permanent dentition with pathognomonic signs of DI (Fig. 2B). The final diagnosis was DI type I.

## DISCUSSION

The dentinogenesis imperfecta has been described in the literature; however, the scientific research in databases of the medical resources, showed little information about this condition.

In these cases, dentinogenesis imperfecta, in association with osteogenesis imperfecta and history of fractures, was evidenced in two of the five brothers, specifically, in cases 2 and 5. In this sense, Trejos *et*

*al.*, reported that the DI type I is always associated with OI, however, only 10-50 % of patients that suffer OI exhibit DI.

The intraoral and radiographic examinations, in association with a right anamnesis, including the history of fractures and family history, allow a presumptive diagnosis of DI and its type. However, for the definitive diagnosis, a genetic study is strictly necessary (de La Dure-Molla *et al.*). Abukabbos & Al-Sineedi stated that DI is caused by mutations in genes that encode type I collagen causing a disturbance in the bone and dentin formation. Majorana *et al.* (2010) added that the temporary dentition is affected more severely than the permanent, the damage being usually less severe in permanent teeth, and in some cases, almost clinically undetectable. The literature (Barron *et al.*, 2008; Rabassa *et al.*; Devaraju *et al.*) describe some radiographic features of teeth affected by DI: crowns with bullous appearance, which have a very strong constriction at the cervical level, shorter and thin roots, and the reduction or obliteration of the pulp chamber. In addition, the same authors pointed out that these features occur in both temporary and permanent teeth, and that the other tissues show a normal appearance. Majorana *et al.*, stated that the yellow/brown discoloration is more prevalent in primary teeth than gray discoloration. All of these clinical and radiographic features concur with the observations in the cases that presented DI.

Arcos *et al.* (2006) and Trejos *et al.*, reported that DI type II only affects the dentin without showing any bony involvement, and that the primary dentition is severely affected, presenting similar clinical and radiographic features to DI type I. In this sense, these features concur with the findings in cases 1 and 4.

Regarding DI type III, Trejos *et al.*, described that clinically, the teeth are less affected than in types I and II, because it exhibits an enamel with normal appearance and the dentin is extremely thin. Moreover, Rabassa *et al.*, state that, radiographically, the dentin is thin, with pulp chambers, and the root canals give the specific appearance of this type of DI, often presenting multiple pulp exposures and radiolucidity in the apical region, which are features that do not correspond to any of the cases presented. The enamel seems to be normal, but may shear rapidly due to deficient dentin-enamel junction, resulting in dentine attrition and loss of the vertical dimension (Abukabbos & Al-Sineedi).

Soto Llanos & Marín (2000) stated that the primary dentition is more frequently affected and highlights the importance of an early diagnosis for proper treatment, maintaining or recovering of the function, and aesthetics. Abukabbos & Al-Sineedi suggest a comprehensive management of these cases with a continuous follow-up and observation, promoting oral hygiene instructions and dietary consultation. The literature (Sánchez Ysmayel, 2000; Vera Cabrera *et al.*, 2003; Abukabbos & Al-Sineedi) describes various treatment alternatives that depend on the characteristics and needs of each case, including the use of stainless-steel crowns to prevent excessive attrition of the affected posterior teeth in the primary dentition, where aesthetics is not as important, while in permanent dentitions affected, an aesthetic and functional rehabilitation treatment is necessary (Goud & Deshpande, 2011; Biethman *et al.*, 2014).

Finally, it is important to know the features of dentinogenesis imperfecta to perform a comprehensive dental care, including the right diagnosis and an effective treatment plan.

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**RESUMEN:** La dentinogénesis imperfecta (DI) es un tipo de displasia de la dentina que afecta su estructura en una o ambas denticiones. La DI puede clasificarse en tres tipos. El objetivo de este informe fue demostrar las características clínicas y radiológicas de los cuatro casos de DI en un mismo grupo familiar. Cinco hermanos fueron controlados clínica y radiográficamente. Dos individuos fueron diagnosticados, por sus características fenotípicas y antecedentes clínicos, con el tipo de DI I; dos de ellos con DI de tipo II y un caso sin signos de DI. Es importante conocer las características de la dentinogénesis imperfecta para poder realizar una atención odontológica integral, lo que permitirá desarrollar un diagnóstico correcto y un plan de tratamiento efectivo.

**PALABRAS CLAVE:** dentinogénesis imperfecta, alteración dentaria, familia, trastornos del desarrollo dental.

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## REFERENCES

- Abukabbos, H. & Al-Sineedi, F. Clinical manifestations and dental management of dentinogenesis imperfecta associated with osteogenesis imperfecta: Case report. *Saudi Dent. J.*, 25(4):159-65, 2013.
- Arcos, H. D.; Yamamoto, A. & Trejo, P. Dentinogénesis imperfecta: Reporte de un caso. *Rev. Odontol. Mex.*, 10(4):173-80, 2006.
- Barron, M. J.; McDonnell, S. T.; Mackie, I. & Dixon, M. J. Hereditary dentine disorders: dentinogenesis imperfecta and dentine dysplasia. *Orphanet J. Rare Dis.*, 3:31, 2008.
- Biethman, R.; Capati, L. R. & Eldger, N. Dentinogenesis imperfecta: a case report of comprehensive treatment for a teenager. *Gen. Dent.*, 62(4):e18-21, 2014.
- Borie, E. E.; Oporto, V. G. & Aracena, R. D. Dens evaginatus in hemophilic patient. A case report. *Int. J. Morphol.*, 28(2):375-8, 2010.
- de La Dure-Molla, M.; Philippe Fournier, B. & Berdal, A. Isolated dentinogenesis imperfecta and dentin dysplasia: revision of the classification. *Eur. J. Hum. Genet.*, 23(4):445-51, 2015.
- Devaraju, D.; Devi, B. Y.; Vasudevan, V. & Manjunath, V. Dentinogenesis imperfecta type I: A case report with literature review on nomenclature system. *J. Oral Maxillofac. Pathol.*, 18(Suppl. 1):S131-4, 2014.
- Goud, A. & Deshpande, S. Prosthodontic rehabilitation of dentinogenesis imperfecta. *Contemp. Clin. Dent.*, 2(2):138-41, 2011.
- Kim, J. W. & Simmer, J. P. Hereditary dentin defects. *J. Dent. Res.*, 86(5):392-9, 2007.
- Majorana, A.; Bardellini, E.; Brunelli, P. C.; Lacaíta, M.; Cazzolla, A. P. & Favia, G. Dentinogenesis imperfecta in children with osteogenesis imperfecta: a clinical and ultrastructural study. *Int. J. Paediatr. Dent.*, 20(2):112-8, 2010.
- Martín-González, J.; Sánchez-Domínguez, B.; Tarilonte-Delgado, M. L.; Catellanos-Cosano, L.; Llamas-Carreras, J. M.; López-Frías, F. J. & Segura-Egea, J. J. Anomalías y displasias dentarias de origen genético-hereditario. *Av. Odontostomatol.*, 28(6):287-301, 2012.
- Min, B.; Song, J. S.; Lee, J. H.; Choi, B. J.; Kim, K. M. & Kim, S. O. Multiple teeth fractures in dentinogenesis imperfecta: a case report. *J. Clin. Pediatr. Dent.*, 38(4):362-5, 2014.
- Rabassa, J.; Palma, C. & González, Y. Dentinogénesis imperfecta?: a propósito de un caso. *Odontol. Pediatr. (Lima)*, 10(2):131-9, 2011.
- Sánchez Ysmayel, A. E. Tratamiento protodóntico en paciente con dentinogénesis imperfecta. Reporte de un caso. *Acta Odontol. Venez.*, 38(2):49-55, 2000.
- Shields, E. D.; Bixler, D. & el-Kafrawy, A. M. A proposed classification for heritable human dentine defects with a description of a new entity. *Arch. Oral Biol.*, 18(4):543-53, 1973.
- Soto Llanos, L. & Marín G., P. Dentinogenesis imperfecta asociada a osteogenesis imperfecta (Tipo I de Shields). Caso clínico. *Rev. Estomatol.*, 9(1):61-7, 2000.
- Surendra, P.; Shah, R.; N. M. R. & Reddy, V. V. Dentinogenesis Imperfecta : A Family which was Affected for Over Three Generations. *J. Clin. Diagn. Res.*, 7(8):1808-11, 2013.
- Trejos, P.; Hernando, V. & De León, C. Dentinogénesis imperfecta: reporte de un caso. *Rev. Estomatol.*, 15(2):19-27, 2007.
- Vera Cabrera, A.; Bustos Alvarez, I. & Castro Barahona, R. Rehabilitación protésica en odontopediatría: Reporte de tres casos clínicos. *Rev. Dent. Chile*, 94(3):13-7, 2003.

Correspondence to:  
Evelyn Borie Echevarría  
Escuela de Odontología  
Universidad Mayor sede Temuco  
Av. Alemania 0281, Temuco  
CHILE

Email: evelyn.borie@umayor.cl

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