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A rare genetic cause of translucent teeth

Una causa genética rara de dientes translúcidos

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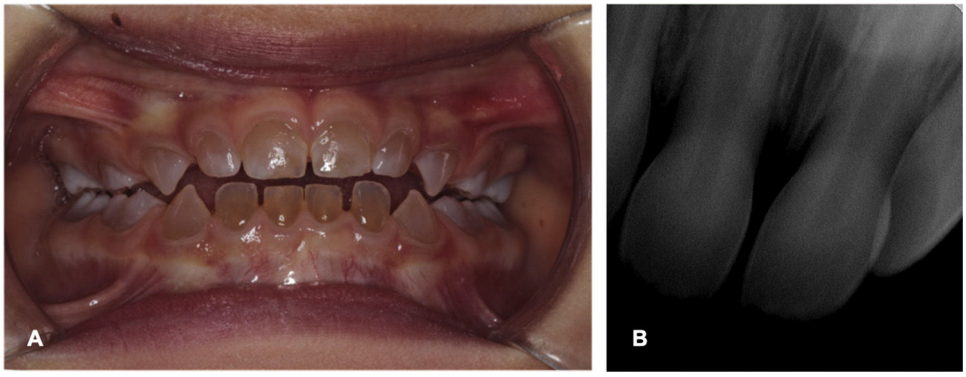

Figure 1 Clinical examination and radiography. (A) Clinical examination of the patient at age 27 months with complete deciduous dentition. (B) Periapical radiographs revealing obliteration of two-thirds of the pulp chambers.

Dentinogenesis imperfecta (DI) is a rare autosomal dominant hereditary dentin disorder characterized by tooth discolouration and translucency in both the deciduous and permanent teeth.[1] Previous studies suggest that DI affects approximately 1 in every 6000–8000 individuals.[2] To date, the dentin sialophosphoprotein (*DSPP*) gene is the only gene with variants known to cause DI, with over 50 heterozygous changes identified to date.[3]

We present the case of a male infant aged 7 months referred by his outpatient care paediatrician to a paediatric dentistry clinic due to the yellowish discoloration and amber-hued translucency of his deciduous teeth (Fig. 1 A). Periapical radiographs revealed obliteration of two-thirds of the pulp chambers, with no discernible changes in enamel thickness, crown structure or root size (Fig. 1 B). A review of

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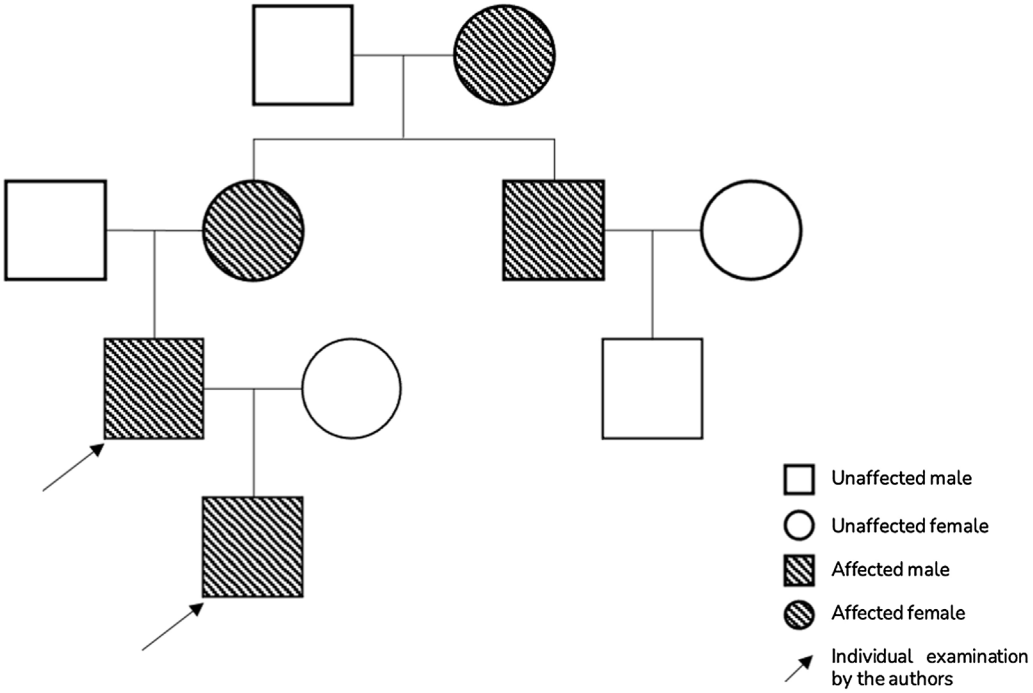

Figure 2 Pedigree of dentinogenesis imperfecta in the family of the patient based on medical records, spanning 4 generations.

the patient's family history revealed similar dental anomalies in his father, paternal grandmother, paternal great-uncle and paternal great-grandmother (Fig. 2). Sequencing of the *DSPP* gene in the patient identified a novel likely pathogenic variant [c.3047del p.(Ser1016llefs*298)] that may explain the presented clinical case.

In cases of genetic dental anomalies, early diagnosis and referral to a paediatric dentist are crucial to prevent early deterioration and loss of teeth.[2] This case report underscores the vital role of paediatricians in recognizing the early signs of the often-overlooked dental alterations that can affect a child's physical and psychosocial well-being.

References

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