Dentinogenesis Imperfecta – “A Hereditary Developmental Disturbance of Dentin”

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Citation

Abstract
Dentinogenesis Imperfecta (DGI) is a genetic disorder of tooth development inherited in an autosomal, dominant way, characterized by the presence of translucent or opalescent dentin, resulting in discoloration (amber-like, bluish grey color) of the teeth. It is a mesodermal defect wherein the dental papilla of either or both the primary and secondary dentition is abnormal. This paper describes the dental findings in a family where the father (33 year old) and the son (3 year old) both showed the characteristic dental features of DGI I.

INTRODUCTION
Dentinogenesis Imperfecta is commonly known as hereditary opalescent dentin or Capdepont dysplasia\(^1\). It is a localized mesodermal dysplasia affecting both primary and permanent dentition and is a genetic disorder of tooth development inherited in an autosomal dominant pattern caused by mutations in the Dentin Sialophosphoprotein (DSPP) gene (cytogenetic location of 4q21.3). The term ‘dentinogenesis imperfecta’ was coined by Robert and Schour in 1939 and probably first recognized by Barret in 1882\(^2\).

Mutations in the DSPP gene lead to the production of abnormal DSPP-derived proteins or reduce the amount of these proteins in developing teeth. As a result, teeth have abnormally soft dentin. Teeth with defective dentin are discolored, weak, and prone to breakage and decay.

The frequency of occurrence of this genetic dental disorder is 1 in 6000-8000 births\(^1\).

Extensive studies have proven over the period of time that DGI is clearly a disorder distinct from Osteogenesis Imperfecta (OI) and hence a revised classification is commonly accepted wherein DGI is classified as DGI 1 and DGI 2.

DGI 1 corresponds to DGI type II and DGI 2 corresponds to DGI type III of Shields classification, respectively. There is no substitute in the revised classification for the category designated as DGI type I of the Shields classification. DGI was earlier classified by Shields et al into three types: \(^3,4\)

Type I, DGI associated with Osteogenesis Imperfecta (OI). Both are mesodermal defects, although OI may occur without DGI.

Type II, that represents DGI without OI

Type III (Brandywine type), that represents a rare variety characterized by shell teeth, with little dentin and multiple pulp exposures in the primary teeth.

The Shields classification appears to be losing ground as it does not account for the molecular aetiologies of the hereditary dentine defects elucidated so far, for example, those underlying Osteogenesis Imperfecta and other syndromes manifesting defective dentine formation.

In DGI, clinically the teeth are discolored into amber-like color, grayish-yellow-brown and possess characteristic opalisation. The crowns take bell-like or bulging shape, the roots are shortened and disfigured. The teeth, shortly after coming out, undergo pathological abrasion\(^5,7\).

As the enamel tends to crack away from the abnormal dentin leaving it exposed and prone to attrition, the patient experiences sensitivity and difficulty in chewing food indirectly impacting the overall health.
Histological structure in Dentinogenesis Imperfecta emphasizes the fact that this is purely a mesodermal disturbance. The enamel layer is thin, but composed properly. The dentine in the external part shows proper structure however in the para-ventricular part it is improper with the canals of a disturbed shape and course; also it has got irregular inter-spherical spaces. The reason for teeth abrasion is the improper structure of dentine and enamel-dentine junction\[8-10\]. Radiographically, the teeth have bulbous crowns that constrict at the cervical region and short roots. Pulp chambers often appear obliterated. It should be noted that periapical radiolucent areas may be visible radiographically, in the absence of any clinically obvious pathology.

CASE REPORT

A 33 year old man reported to the department of Oral Medicine and Radiology at PDM Dental College and Research Institute, Bahadurgarh, Haryana, India with the chief complaint of his child, aged 3 years, having small sized discolored teeth since birth and hence desired them to be restored. The medical examination (including blood tests and full body X-rays) of the child did not reveal any systemic abnormality or specifically symptomatic of Osteogenesis Imperfecta. Interview and clinical examination of the father revealed similarly discolored and small sized teeth since birth (Fig 1, 2). Further, the family history of the child showed absence of disease in mother and sibling (sister). Intra oral examination of both the child and his father showed all teeth having amber like discoloration (Fig 3). None of the child’s teeth showed mobility or tenderness on percussion, however, percussion test of the father’s teeth revealed tenderness in relation to 36. Both the father and the child’s maxillary and mandibular teeth showed complete chipping of enamel with exposure of underlying yellow dentin. Further, generalized attrition was also noted, specially, buccal-cusp attrition with the primary molars in the child (Fig 1, 2, 4). No soft tissue findings were noted. Radiographic findings of the child showed smaller crown size and complete obliteration of the pulp canals (Fig 5).

OPG of the patient’s father revealed smaller sized crowns, bulbous roots, prominent cervical constriction along with complete obliteration of root canals in all permanent teeth. Periapical radiolucency was seen in relation to 36 (Fig 6). Due to its non restorable condition 36 was extracted and its ground sections were made. The histological findings showed irregular sparse dentinal tubules. Some areas also showed complete absence of dentinal tubules. Reduced pulpal space was also noted in the ground sections of 36 (Fig 7).

DISCUSSION

The greatest challenges faced in the treatment of patients with Dentinogenesis Imperfecta were extreme fragility of the teeth, doubtful retention of the esthetically sound restorations. Such patients usually present with the complaint of unesthetic teeth, pain, chronic dental abscess, and/or decreased vertical height. Early diagnosis and treatment intervention helps in better prognosis and rehabilitation in such cases. According to Wei, stainless steel crowns are commonly used to prevent the attrition of the deciduous teeth and young permanent teeth\[11\]. The use of potentially abrasive materials in opposition to unrestored tooth surfaces should be avoided. The importance of implementation of early preventive measures in order to avoid pulpal pathology from occurring has been demonstrated, as any necessary ensuing endodontic therapy would be difficult to execute and any endodontic treatment provided has a poor prognosis.

Depending upon the economic status of the parents, composite veneers or celluloid crowns can be chosen. In a study by Harley and Ibbetson\[12\], the longevity of adhesive castings in patients affected by amelogenesis and dentinogenesis imperfecta was examined .It was found that despite the abnormally formed tooth structure in these patients only one casting was lost out of a total of sixty four which had been placed in at time period of between ten and fifty four months.

In the study, two patients who suffered with DGI had nickel-chromium adhesive castings placed on their anterior teeth. The casting remained in situ for 16 and 17 months, respectively. Thus, although the numbers studied were small, the value of using adhesive castings to protect underlying tooth tissue and restore aesthetics in such patients should not be underestimated\[12\].

The permanent teeth can be restored with porcelain jacket crowns, fixed or removable prosthesis, overdentures or implants. Shafer et al emphasize on the doubtful retention of restorations due to diminished quantity and quality of dentin present\[13\].

Owing to risk of enamel fracture, intracoronal restorations are contraindicated. The shortened root length means that crown lengthening procedures should be avoided or
performed with caution.

In this case the authors aimed at providing treatment, along with preventive measures, to achieve a good esthetic result and restore the function.

**Figure 1**
Figure 1: Intra oral (maxilla) clinical presentation of the father

**Figure 2**
Figure 2: Intra oral (mandible) clinical presentation of the father

**Figure 3**
Figure 3: Intraoral presentation of the child

**Figure 4**
Figure 4: Intraoral presentation of the mandible(child) showing buccal cusp attrition

**Figure 5**
Figure 5: OPG of the child showing the typical features of DGI

**Figure 6**
Figure 6: Orthopantomograph of the father showing the complete obliteration of the pulp chambers, periapical radiolucency was also seen in relation to 36
**Figure 7**

Figure 7: Histopathological section of 36 (father)

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**TREATMENT**

The treatment of the child included oral prophylaxis, delivering of oral hygiene instructions and diet counseling. Composite veneers were placed for maxillary and mandibular anteriors to restore better esthetics. Prefabricated stainless steel crowns were placed for maxillary and mandibular primary molars as prophylactic measure. Beginning in infancy, treatment usually continues into adulthood with a number of options including the use of crowns, over-dentures and dental implants depending on the age of the patient and the condition of the dentition.

However, due to the weakened condition of the teeth, many common cosmetic procedures such as braces and bridges are inappropriate for patients with dentinogenesis imperfecta and are likely to cause even more damage than the situation they were intended to correct. Frequent dental appointments in which dentists provide specific instructions regarding patients’ diets such as avoiding hard foods, as well as frequent prophylaxis and applications of topical fluoride, help prevent carious lesions in adulthood.

Psychological and Genetic counseling of the child and the family holds true importance in these cases. Impacted families should be educated regarding the maintenance of good oral hygiene and regular follow-ups for dental checkups and maintenance. Special counseling over genetic disorders needs to be provided. As DGI is inherited in an autosomal dominant fashion, there is a 50% chance that a child born to an affected parent will also be affected. It is imperative to emphasize on prenatal examinations that help in proactive determination of such genetic disorders. Where diagnosis occurs early in life and treatment follows the outlined recommendations, detrimental effects on the developing dentition can be prevented.

**References**

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