A case report of dentinogenesis imperfecta inherited in a family of four generations
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Abstract
In dental anomaly, dentinogenesis imperfecta (DI) is one of the hereditary developmental disorders affecting both formation and maturation of dentin. This autosomal dominant disorder is characterized by discolored and translucent teeth ranging from gray to brownish-blue or amber. It can affect both primary and permanent teeth. The enamel may split readily from the dentin on occlusal stress. Radiographically, there is evidence of cervical constrictions, short root, and pulp chambers. The affected teeth may have smaller or completely obliterated root canals. This article reports a case of a family, which is affected by type I DI over four generations.

Keywords: Amber colored teeth, bulbous teeth with cervical constrictions, dentinogenesis imperfecta, hereditary

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Introduction
Dentin defects, with hereditary predisposition, are rare diseases that affect primarily the organic dentin matrix. Dentinogenesis imperfecta (DI) is one among them which is classified into three types: DI-I, DI-II, and DI-III. All three types are inherited by an autosomal dominant transmission that affects both deciduous and permanent teeth.[1] The classifications given by Witkop and Shields are generally accepted, but they are not satisfactory. Genetic researches on the Type I DI have been confirmed that the condition of osteogenesis imperfecta is a separate pathology from DI. However, it is showed that both Type II and III of DI are caused by a single mutation of gene named dentin sialophosphoprotein. Neville introduced a new classification after incorporating the knowledge generated by recent research.[2] We are reporting a case of DI Type I, in which, four generations of a family of Bengaluru city, Karnataka, India, are affected by the disease.

Case Report
A patient (male; 35 years) reported to Oral Medicine and Radiology Department with a complaint of decayed upper right and lower left back teeth since a year, at Sri Rajiv Gandhi College of Dental Sciences and Hospital, Bengaluru, Karnataka. He had experienced moderate pain and swelling in the same teeth region a few weeks back, which subsided on medication. No significant medical history and allergy history were reported. Past dental history revealed he had discolored teeth from the time his permanent teeth erupted [Figures 1 and 2] and also reported that his deciduous teeth were bluish in color. On further questioning about family history, the patient revealed that his mother [Figure 3] aged about 62 years, and her sister also has teeth discolorations. His grandmother also reported to have the same type of teeth discolorations. Mother and her sister were having reddish hue in their teeth. Patient’s sister was also having reddish hued teeth. His mother’s sister has three children, and all of their teeth are discolored. The patient gave history of consanguineous marriage (with his mother’s younger sister’s daughter aged 30 years). His wife also has discolored teeth [Figure 4]. They have two children, among them; the younger, female child has discolored teeth [Figure 5]. Intraoral examination of the patient showed amber colored discoloration of all teeth. The patient has generalized attrition of teeth, root stumps in relation to teeth number 16, 14, 36. The ceramic bridge was placed in relation to 25, 26, 27 and 44, 45, 46, 47, 48. Mobility was seen in relation to 11, 12, 21, 22 [Figure 2]. The patient was advised for an intraoralperiapical radiograph and orthopantomograph (OPG), which showed generalized haziness of enamel, bulbous crowns with constriction at the cervical aspect of teeth, generalized horizontal bone loss up to cervical third of roots, and generalized obliteration of pulp cavity and root canals [Figure 6]. Figure 6 shows OPG of the patient showing...
Figure 1: Intraoral view of patient showing the typical color deviation and unusual translucency

Figure 2: Extensive wear of posterior teeth, missing teeth, ceramic bridge over the teeth

Figure 3: Patient’s mother teeth with the same discolorations and destructed teeth

Figure 4: Patient’s wife teeth with discolorations and translucency

Figure 5: Patient’s daughter with the newly erupted deciduous teeth showing discoloration

Figure 6: Orthopantomograph of patient showing generalized haziness of enamel, bulbous crowns with constriction of cervical aspect of teeth.

generalized haziness of enamel, bulbous crowns with constriction of cervical aspect of teeth. The differential diagnosis for DI is dentin dysplasia. However, a premature teeth loss, severe teeth attrition, bulbous crowns, marked cervical constrictions and few periapical radiolucencies are the classical features of DI, differentiating it from
dentin dysplasia. Based on above mentioned clinical presentations and radiographic findings, the case was diagnosed as DI. The patient was advised for extraction of grossly destructed teeth. Those extracted teeth were sent to the Department of Oral Pathology of the same institution for ground sectioning and staining for confirmation of diagnosis. Ground section of the teeth showed a thin layer of surface enamel with scalloped dentinoenamel junction in only a few areas, hypocalcification with obliteration of pulp was seen. H and E staining showed irregular tubular arrangement and granularity and vascular spaces in the dentin matrix confirming the case as DI. Root canal treatment was done for few teeth and replaced with ceramic crowns.

Discussion

DI is a hereditary disorder of dentin formation. Global statistics reports that 1 in of 8000 individuals suffers from this disorder. In 1939, Robert and Schour coined the term “DI.” Earlier, Shields had divided this condition into three types of dentin defects (DI Type I, II, and III). Witkop considered Type I defects as defects as DI; Type II as hereditary opalescent dentin, and III as Brandywine isolate, respectively. However, the recent genetic studies led to a newer classification. However, Barrett was pioneer to recognize this disease during last part of the 19th century. Talbot for the first time, in the year 1893, published a report describing this condition as a defect of enamel. In 1908, Fargin-Foyelle along with Malassez recognized that the defect is because of abnormal dentin. Teeth with the DI show a mesodermal defect, in which there is an abnormality in the primary structure of dentine. Studies by Kerebel et al. and Wright et al. showed abnormalities in the dentinal tubules and calcification of dentine in such teeth. No changes were seen in enamel, cementum, and periodontal ligament of affected teeth. Microfractures of dentin and defects in dentinoenamel junction cause chipping of teeth. A transparent dentin in DI is because of greatly reduced or completely absent dentinal tubules. The dentin affected by DI is observed to have reduced amounts of calcium, phosphorus, and magnesium. It also shows an abnormally higher ratio of calcium and phosphorous. A higher water content in such teeth is also a general feature. Higher attrition, a common clinical presentation of DI-I, is the result of intratubular mineralization. Premature fracture is the implication of decreased mineral concentration in dentin. In the presented case, the family pedigree [Diagram 1] revealed that an affected couple gave birth to an affected child. This shows the genetic involvement of the disease. Diagram 1 shows pedigree appropriate treatment after timely diagnosis of DI may prevent morbidity to the patient with respect to psychological and functional aspects. With advancing age, providing an optimal treatment to the patient is diminished. Most of the patients suffering from DI require an interdisciplinary treatment strategy. A dentist should determine an appropriate treatment considering the age, clinical presentation, extent of suffering, expectations, and resources available with the patient.

 Conclusion

Since the patients of DI present with varied severity, a dental practitioner must be equipped with the knowledge to diagnose and design an appropriate, multidisciplinary treatment plan to rehabilitate them. This report is an attempt to create awareness about DI among the dental surgeons to educate common masses on this genetic oral disease.

References