**ABSTRACT**

Dentinogenesis imperfecta (DI) is a developmental tooth disorder affecting dentin, characterized by the presence of opalescent dentin caused due to autosomal dominant pattern resulting in a dusky blue to brownish discoloration of the teeth. It can affect both deciduous as well as a permanent dentition. It can be classified as DI-I and DI-II. This case report describes a 16-year-old male patient with clinical and radiographical characteristic features of DI-I. A brief note on etiology, differential diagnosis and treatment plan will be reviewed.

**Keywords:** Autosomal dominant, Dentin, Dentine hypoplasia, Dentinogenesis imperfecta, Mutations

**INTRODUCTION**

Human dentition can subject to numerous variations in terms of size, shape, number or change in structure during formation of dental tissues. Dentinogenesis imperfecta (DI) is one such disorder, which affects dentine.1,2

It was probably first recognized by Barret in 1882. The first published report describing the disorder as an enamel defect was by Talbot as quoted by Witkop.3 The term “hereditary opalescent dentin” was first used by Skillen,4 Finn5 and Hodges6 to describe the brown translucent teeth, which are opalescent, lacking pulp chambers.

DI is a localized mesodermal dysplasia affecting both the primary and permanent dentitions, in an autosomal dominant fashion with high penetrance and a low mutation rate.7 It is the most common dental genetic disease, affecting approximately 1 in 8000 births.8

The color of the teeth varies from brown to blue and is sometimes described as amber or gray. Enamel may show hypoplastic or hypocalcified defects in about one-third of the patient’s and in an affected person tends to crack away from the defective dentin. The exposed dentin may then undergo severe and rapid attrition.7 The underlying defect of mineralization often results in shearing of the overlying enamel leaving exposed weakened dentine, which is prone to wear.

The classification system for DI was proposed by Shield as - type 1, type 2 and type 3. However, shield’s classification had certain drawbacks, so it was revised into DI-I and DI-II. Recent genetic studies have shown that mutations in the genes encoding collagen underlie this condition.

Diagnosis is based on family history and detailed clinical examination, while genetic diagnosis may become useful in the future once sufficient disease causing mutations have been discovered. Differential diagnosis include hypocalcified forms of amelogenesis imperfecta, congenital erythropoietic porphyria, conditions leading to early tooth loss (Kostmann’s disease, cyclic neutropenia, Chediak-Hegashi syndrome, histiocytosis X, Papillon-Lefevre syndrome), permanent teeth discoloration due to tetracyclines, vitamin D-dependent and vitamin D-resistant rickets.

Radiographically, the teeth have bulbous crowns, constricted short roots. Initially, the pulp chambers may be abnormally wide, giving the appearance of “shell teeth,” but they progressively get obliterated.

Treatment involves removal of sources of infection or pain, improvement of esthetics and protection of...
the posterior teeth from wear. Beginning in infancy, treatment usually continues into adulthood with a number of options including the use of crowns, overdentures and dental implants depending on the age of the patient and the condition of the dentition. If the diagnosis occurs early in life, treatment options are ample with good prognosis.9

**CASE REPORT**

A 16-year-old male patient, visited to the Department of Oral Medicine and Radiology, with a complaint of tooth chipping and yellowish to brown teeth, since the time permanent teeth erupted, but deciduous teeth were normal. Furthermore, the patient had increased sensitivity to cold water and food items. Family history revealed patient’s father had similar yellowish brown teeth.

On clinical examination, generalized teeth appeared yellowish brown in color and translucent, attrition present and tooth surface chips off (Fig. 1). Dentinal caries was approximating pulp and tenderness present on vertical percussion irt 16 and 26. Clinically missing teeth were 18, 28, 38 and 48. Occlusion is skeletal Class III malocclusion.

Hence, based on the patients history and clinical findings, provisional diagnosis given was DI (generalized).

Patient’s Full mouth Intra-oral periapical radiographs (Fig. 2) and orthopantomogram (Fig. 3) revealed generalized obliteration of coronal and radicular pulp chamber. Crown appeared bulb shaped with constricted cervical areas. Roots were short and narrow. Cementum, alveolar bone and periodontal ligament space were normal. Hand wrist radiograph and anteroposterior view were taken to rule out DI associated with osteogenesis imperfecta and no significant findings were detected in the resultant radiograph, and it was ruled out. Thus, final diagnosis of DI-I - Generalized, according to revised classification was given.

Patient advised to undergo generalized prosthetic rehabilitation and orthodontic evaluation for correction of malocclusion.

**DISCUSSION**

According to revised classification DI-I is a disease inherited in a simple autosomal dominant mode. The classification system of hereditary disorder was first formulated by Shields in 1973 into three types of DI-I, II, III.9 However, Shields system of classification does not describe the molecular etiologies of the hereditary dentin defects. Hence the revised classification:

- DI-I which is not associated with osteogenesis imperfecta. This corresponds to DI type 2 of shield’s classification.
- DI-II which is associated with osteogenesis imperfecta. This corresponds to DI-III of shield’s classification. There is no substitute in the present classification for the category designated as DGI type 1 of the previous Shields classification.10

Genetically DI-I is transmitted as an autosomal dominant Mendelian trait with almost 100% penetrance. It probably represents a basic defect in structural or regulatory protein and the defective gene has been identified as dentin sialophosphoprotein and has been mapped to chromosome 4q21.3. The gene product is a precursor protein that is cleaved into 2 dentine specific proteins, dentine sialoprotein and dentine phosphoprotein.11

Clinically, the appearance of the teeth with DI is characteristic. They show a high degree of amber like translucency and a variety of colors from yellow to blue-gray. The colors change according to whether the teeth are observed by transmitted light or reflected light.12,13 Affected teeth have broad crowns with constriction of the cervical area resulting in a tulip shape.12 The enamel easily fractures from the teeth and the crowns wear readily. In adults, they may frequently wear down to the gingiva. The exposed dentin becomes stained. The color of the abraded teeth may change to dark brown or even black. Some patients demonstrate an anterior open bite.13

Radiographically the teeth appear solid, lacking pulp chambers and root canals.14 Radiographs may also reveal slight to marked attrition of the occlusal surface. The roots are usually short and slender.

Thus the diagnosis is based on history, clinical examination and radiographic features.
The conditions included in the differential diagnosis have similar clinical or radiographic features of DI-I. They can be divided under three categories.

I. Clinical
II. Radiographic
III. Clinical and Radiographic

Clinical

Exposure of underlying dentine

Hypocalcified forms of amelogenesis imperfecta initially develop normal enamel thickness, but the poorly calcified enamel, is soft and friable and is rapidly lost by attrition leaving dentine cores. Unlike in DI-I, the teeth are usually sensitive and on radiographs enamel is less radio-dense than the dentine.

Intrinsic discoloration

Deposition of red-brown pigments in the bones and teeth is seen in congenital erythropoietic porphyria resulting from an inborn error of porphyrin metabolism. A number of prenatal and neonatal enamel discolorations and hypoplasias are due to neonatal hemolytic anemia’s. The discoloration, which ranges from yellow to green, brown and grey to black is usually found at the necks of teeth, and the enamel hypoplasias are usually located in the coronal third of the teeth.

Tetracycline stains vary from yellow or grey to brown depending on the dose or the type of the drug received in relation to body weight, but in DI-I teeth appears yellow or blue gray.

Radiographic

Regional odontodysplasia is of unknown etiology. Radiographically, roots are short with wide open apices and very wide pulp canals. Primary and permanent teeth are affected. Irradiation to jaws or chemotherapy during the period of root development leads to arrested development and can give a radiographic appearance of DI.

Clinical and Radiographic

Vitamin D-dependent rickets and vitamin D-resistant rickets have clinical and radiographic features of DI. Vitamin D-dependent rickets is characterized by yellowish to brown enamel, chronic periodontal disease, large quadrangular pulp chambers and short roots. Features of vitamin D-resistant rickets include attrition and exposure of abnormally formed dentine of primary teeth and abscessed non-carious primary or permanent teeth.

In DI, early in development, the teeth may appear to have large pulp chambers, but these are quickly obliterated by the formation of dentin. Ultimately the root canals may be absent or threadlike. Occasional periapical radiolucencies are seen in association with sound teeth without evidence of pulpal involvement, which may occur from microscopic communication between residual pulp and the oral cavity. These lesions do not occur as frequently as in dentin dysplasia. The architecture of the bone in the maxilla and mandible is normal.

Histologically the dentin is composed of irregular tubules, often with large areas of the uncalcified matrix.
The tubules tend to be larger in diameter and less numerous in a given volume of dentin than in normal teeth.\textsuperscript{14}

There are no guidelines on restorative treatment in teeth affected by DI. The treatment should begin as early as possible, considering the degree of tooth destruction and patient cooperation.\textsuperscript{21}

Wright has stated that the dental approach for managing DI will vary with the severity of the clinical expression while intracoronal restoration and bonded veneers for anterior teeth may be acceptable in mild cases; they might not last in severe cases exhibiting enamel fracturing and rapid wear.\textsuperscript{22}

The objectives of early treatment of DI in the primary dentition are as follows:

1. Maintain dental health and preserve vitality, form, and size of the dentition.
2. Provide the patient with an esthetic appearance at an early age, in order to prevent psychological problems.
3. Provide the patient with a functional dentition.
4. Prevent loss of vertical dimension.
5. Maintain arch length.
6. Avoid interfering with the eruption of the remaining permanent teeth.
7. Allow normal growth of the facial bones and temporomandibular joint.
8. Establish a rapport with the patient and the patient’s family early in the treatment.\textsuperscript{23}

Treatment of the mixed and permanent dentition is challenging and frequently demands a multidisciplinary approach. Collaboration of the pediatric dentist with a prosthodontist and an orthodontist is often imperative. Although caries is not a major concern in most cases, strict oral hygiene instructions and preventive treatment is important in order to prevent caries from adding to existing problems. Frequently, there is a need to reestablish the vertical dimension of occlusion in order to restore the occlusion in the mixed and permanent dentition. Prosthetic restoration combined with orthodontic treatment may be advantageous, and evaluation of the occlusion prior to initiation of treatment is advised.\textsuperscript{24}

**CONCLUSION**

The greatest challenge to the dental surgeon is the total rehabilitation of the patient with DI as it requires the active involvement of various branches of dentistry and it is important that this type of condition should be facing towards functional and esthetic rehabilitation. Thus overall improvement of esthetic effect, which in itself can also contribute to patient’s psychosocial status.

**REFERENCES**


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