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Review

DENTINOGENESIS IMPERFECTA: A REVIEW

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ABSTRACT

Dentinogenesis imperfecta (DGI) is an inherited dentin disease resulting in fragile teeth that affect the primary and permanent dentitions. It causes physical and aesthetic problems, including tooth discoloration and staining, and is responsible for great damage and wear to the entire dentition. DGI is classified into three types, with COL1A1 and COL1A2 gene mutations present in the first type, and mutations of the DSPP gene present in the second and third types. Treatment is focused on the restoration and replacement of damaged and worn teeth, as well as aesthetic improvements. Restorative, prosthodontic, and orthodontic treatment is often necessary. The aim of this paper is to shed light on this pathological disease and the importance of obtaining an early diagnosis which is necessary to implement treatment.

KEYWORDS: *DGI, dentin defect, dental, musculoskeletal*

INTRODUCTION

Dentinogenesis imperfecta (DGI) is an autosomal dominant inherited dentin disorder that affects both the primary and permanent dentitions. It causes physical and aesthetic problems, including opalescent dentin, brown or blue-hued tooth discoloration and staining, and is a common genetic dentin defect, estimated to affect 1 in 8,000 people (1).

A brief look at the history of DGI shows that it may have been first identified in 1882 by Barret, and that the enamel deformity occurring in the disorder was first described in literature in 1971 (2).

DGI was originally classified in three types in 1973 by Shields; these being Type I, II and III (3). These are continued to be referenced today, as well as a newer, revised system of two types.

This review outlines the current literature on DGI with a summary of the clinical characteristics, the classification of the disorder into subgroups, and the standard treatment options. Because of the serious physical and aesthetic problems associated with the disorder, an early diagnosis is vital.

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Clinical description

DGI is an autosomal dominant disease consisting of dentin defects, altered dentin mineralization and limited root dentin. The teeth are made up of four different tissues, which include the dentin, enamel, cementum, and pulp. The teeth are mostly composed of dentin, which is softer than enamel (4). This dentin is altered in people with DGI.

DGI is characterized by opalescent dentin, a characteristic discoloration and staining of the teeth, enamel loss, tooth weakening and erosion, bulbous crowns, cervical restriction, short roots, and destructed root canals and pulp chambers (5). Clinical severity of the disease is variable and can range from mild to very severe (6).

Crown discoloration is a pronounced characteristic, and can present with grey, bluish-grey, and amber-brown tinges (7). Teeth present with a translucent, opalescent quality. These aspects are aesthetically displeasing and can cause phycological distress for the patients.

Structurally, the teeth are very delicate and wear easily. Enamel damage, chipping and crown destruction can present, and the crowns themselves may appear as bulbous or domical and smaller than usual (8). Cervical restriction due to the erosion of the hard tissues around the neck of the teeth may also be noted. The roots are shortened, and canals and pulp chambers can be destructed or obliterated (9).

Diagnosis of DGI is usually obtained through clinical examination, radiography, and assessments of family history, and genetic counseling can be confirmative.

A case revision was presented by de La Dure-Molla which compared the severity of clinical features between the primary and permanent dentitions. This showed that milder symptoms occurred in the cases of the permanent teeth, with milder and severe symptoms presenting in the cases of the primary teeth (10).

Classification

DGI was classified into three types by Shields. All three types are inherited genetic disorders, but the responsible genes vary.

Type I DGI is associated with osteogenesis imperfecta, a congenital disorder linked to mutations of the COL1A1 or COL1A2 genes, that leads to brittle bones and skeletal irregularities. Oral hygiene is affected with tooth loss being common. Between 20 and 40 percent of people suffering from osteogenesis imperfecta may have Type I DGI (11).

Dentin sialophosphoprotein (DSPP) gene mutations are present in DGI Types II and III. The DSPP gene encodes proteins that create the dentin extracellular matrix, which include collagen type I and the non-collagenous proteins responsible for DGI such as dentin sialoprotein (DSP), dentin glycoprotein (DGP) and dentin phosphoprotein (DPP) (12). Mutations of the DSPP gene alter the dentin-producing proteins and are responsible for the dentin defects of the disorder.

Type II DGI, the most common DGI type, shares the same clinical characteristics of Type I, however the distinguishing factor is that it is not associated with osteogenesis imperfecta (13).

Type III DGI is a rare and severe form, associated with a specific U.S. population of Brandywine, located in Southern Maryland near Washington, DC (14). In Type III DGI there is rapid erosion and damage to teeth, with the possibility of dental pulp becoming exposed (15).

Shield's classification of DGI into these three types was clinically based and is still relevant today, however recent genetic research by de La Dure-Molla has shown that the three types are a variation of the same disease, and therefore a revised classification system was created that divides the diseases into two groups of DGI types II and III (10).

Treatment

Early diagnosis of DGI is vital to initiate proper treatment, as the problematic dentin formation of DGI that leads to tooth erosion and fractures, and the destruction can progress quickly, eventually requiring corrective dental procedures (16). Treatment follows a multidisciplinary strategy that depends on the patient's age and severity of the disease (17). Restorative, prosthodontic, and orthodontic treatment is often necessary (18). Improving the aesthetic features of patients is also of great importance in regard to the patient's self-image and oral health-related quality of life (19).

Treatment usually begins with the primary teeth with fillings or crowns. Dental implants and dentures are standard for permanent dentition when replacement becomes the primary option. Orthodontic treatment including removable appliances, functional appliances, and fixed appliances, can be a long process and may require years of treatment, which can result in frustration and difficulty for the patient (20).

Dental implants may have limited success, as they require a solid tooth base to adhere to which may be absent with the damage caused by DGI. There is a risk of implant failure.

Porcelain fused to metal (PFM) crowns and bridges, veneers, as well as stainless steel, composite and all-ceramic crowns are used to treat DGI. Caries are common and pose a great risk of failure for restorations. Therefore, impeccable oral hygiene is of great importance and the patient must commit to this, even in the face of tooth pain and sensitivity (21).

In Type I bisphosphonates are sometimes prescribed to treat the bone problems of the accompanying disorder osteogenesis imperfecta. While these bisphosphonates improve bone qualities, they can negatively impact the oral restoration process (22).

CONCLUSION

Although DGI may not be a prominent hereditary disorder overall, it is the most common genetic dentinal disease. The attrition and destruction to dentition, as well as the aesthetic impacts of discoloration and staining, can have a severe, negative impact on the patient's life. The oral quality of life is greatly affected, and subsequent dental restorations and replacement may demand an extensive amount of time and commitment on the end of the patient. Dental anxiety may form as a result. The patient may also suffer with poor self-image and experience negative social aspects resulting from the aesthetic qualities of their teeth.

Progress can be made by further investigation into the genetic basis of DGI and the classification of the subtypes, important research for early diagnosis and subsequent treatment.

Conflicts of Interest:

The authors declare no conflict of interest.

Author Contribution:

The author has read and agreed to the published version of the manuscript.

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