Osteogenesis Imperfecta

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Abstract

Osteogenesis Imperfecta (OI), also known as glass bone disease; It is an autosomal dominant inherited disease with a prevalence of approximately 1/15,000-1/20,000, with clinical findings such as increased bone fragility, blue sclera, dental disorders, dentinogenesis imperfecta, hearing loss, ligamentous laxity, long bone fractures and deformities. In OI, which is the most common cause of genetically-induced osteoporosis, long bone fractures and vertebral compression fractures are seen after a simple trauma. In OI patients, dental anomalies are seen clinically as dentinogenesis imperfecta (DI), malocclusion, mandibular prognathism, and decrease in vertical dimension. In this article, it is aimed to evaluate an important bone disease such as osteogenesis imperfecta by emphasizing its clinical and oral findings.

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Keywords: Osteogenesis imperfecta, blue sclera, brittle bone disease, dentinogenesis imperfecta.

Introduction

Osteogenesis Imperfecta (OI) is a hereditary connective tissue disorder with a prevalence of approximately 1/15,000-1/20,000, characterized by increased bone fragility and decreased bone mass, also referred to as 'brittle bone disease' or 'glass bone disease'(1, 2). OI type 1 is a disease that occurs as a result of the defect of genes that provide collagen formation (3).

Type 1 collagen; It is a rigid triple helix protein consisting of two pro-1 chains and a pro-2 chain. It has been reported that OI occurs as a result of mutations of genes in both chains of type I collagen, COLIA2 and COLIA1(4). It is the most abundant protein in mammals and is the main protein of bone. Therefore, most clinical manifestations of OI affect bone structures. Since type 1 collagen is also found in tendons, ligaments, sclera, dentin, skin, middle and inner ear, many problems are encountered in patients with OI (4-6).

Diagnosis of OI; It is determined by patient history, clinical and genetic tests, bone biochemistry, lumbar spine bone mineral density (BMD) and radiographic findings. It has been reported that a multidisciplinary treatment approach is required in patients with OI, and skeletal disorders such as bowing of the long bones, curvature of the spine, deformities in the rib cage, and susceptibility to bone fragility are encountered depending on the age and severity of the disease(7, 8). It has been found that blue sclera (the appearance of the underlying vascular layer due to excessive thinning of the sclera in the eye), growth retardation, dental disorders, dentinogenesis imperfecta, abnormal bone formations, thin skin, joint hypermobility, hearing impairment and hair loss are observed in OI patients (2, 9).

The most widely used classification of OI belongs to Silence et al. in 1979 and it has been reported that it is classified into four clinical types according to clinical, radiographic and genetic features (1, 10, 11).

Type 1 OI; It is the most common and mildest type, and the patients have a normal appearance. Type 2 OI with blue sclera, no bending of long bones and hearing impairment; it is the most severe type and multiple bone fractures cause fetal death, in patients with type 3 OI; The scleras are blue at birth and decrease with age, bone deformities that start with birth and progress, triangular face, hearing loss, muscle weakness, bone fractures, and developmental delay are observed, this group of patients are wheelchair bound at a very early age and in type 4 oi patients; It has been reported that there are wide phenotypic differences between mild and severe forms of OI, limitation of mobility, short stature, vertebral fractures, bowing of long bones, and shortened life span (3,5,11-13). (Table 1)

Table 1. OI classification by Silence et al (5).

Type	General manifestations	Specific manifestations
I- Autosomal dominant inheritance with blue sclera.	Variable bone fragility, blue sclera, early deafness, mild stunting.	I-A: normal teeth.
		I-B and I-C: dentinogenesis imperfecta.
II- Perinatal lethal form, radiographically characterized by crumpled femora and beaded ribs.	Extreme bone fragility, perinatal death.	II-A: short and widened long bones with fractures, wide ribs with fractures.
		II-B: short and widened long bones with fractures, ribs with sparse fractures.
		II-C: thin long bones with fractures, thin ribs.
III- Progressively deforming, with normal sclera.	Moderate to severe bone fragility, blue sclera in infancy.	Early-onset kyphoscoliosis.
		Dentinogenesis imperfecta may be present.
IV- Autosomal dominant inheritance with normal sclera.	Bone fragility, moderate to severe deformity of the long bones and spinal column, white sclera, moderate to severe stunting.	IV-A: normal teeth.
		IV-B: dentinogenesis imperfecta.

Oral manifestations of OI are dentinogenesis imperfecta (DI), malocclusion, mandibular prognathism and loss of vertical dimensions. DI is a hereditary disorder characterized by defect in dentin formation in permanent and primary dentition and is divided into 3 groups. DI type 1 is linked with OI, and is caused by mutations affecting collagen formation. Types 2 and 3 DI are caused by mutations in the gene encoding dentine sialophosphoprotein (DSPP). In all 3 forms, the teeth appear opalescent sheen, ranging from brown to blue-gray. Clinically, teeth often manifest wear/loss of enamel due to altered dentine-enamel junction. The radiographic features of DI Types I and II are similar, exhibiting bell-shaped crowns, short blunt roots, dental pulp obliteration and periradicular radiolucencies. In contrast, pulp enlargement giving shell teeth aspect is radyographic feature of DI type III (11, 14, 15).

O'Connell and Marini; showed in their study that the discoloration and attrition in the teeth of DI patients were more severe in the permanent dentition than in the primary dentition. O'Connell and Marini also showed that teeth with DI are more resistant to caries than normal teeth and explained the reason for this is the rapid abrasion of the exposed dentin and the lesser and irregular structure of dentine tubules (11, 16).

It is very important to know the current medical status, to learn the type and severity of the disease. Radiographic examinations of these patients are performed with periapical radiographs. Panoramic radiographs are not obtained due to shortness of the neck, largeness of the chest and mobility impairments. The importance of oral hygiene should be explained to such patients because of the increased risk of fracture (4, 11).

In order to develop appropriate treatment strategies in patients with OI, it is necessary to know their current medical status. Excessive number of decayed teeth and periodontal problems are frequently observed in these patients. OI is related to an increased risk of hypodontia as well as oligodontia. The treatment planning should be aimed to preserve vertical dimension, function, aesthetics, normal growth and development. In addition, conservative treatment approach should be planned to these patients as much as possible. Full coverage restorations or other prosthetic treatment like overdentures may be essential to preserve function and aesthetics. In addition, it was emphasized that orthognathic treatment and selective orthodontic may be beneficial in limiting malocclusion (6). OI patients can use antiresorptive drugs such as bisphosphonatesas they reduce fracture risk and delay progression of the disease. The main mechanism of the drug is inhibiting the

function of osteoclasts, preventing bone resorption and reducing bone pain (17).

It should be said that it is important to carry out oral hygiene and dental preventive dentistry measures in these patients, since poor oral hygiene causes dental complaints.

Since osteogenesis imperfecta is a hereditary and clinically evident disease, these patients come to the dentist with the diagnosis made. However, there may be mild cases that are overlooked. It should not be forgotten that osteogenesis imperfecta may coexist in patients who are suspected of having dentinogenesis imperfecta after oral clinical and radiological examination, and the physician should definitely take a more detailed anamnesis.. If there is a positive family history, bone pain, bone fracture tendency, or hearing problem as a result of the anamnesis, the patient should be referred to a specialist physician with multidisciplinary approaches for detailed examination. Regular dental examinations should be performed in patients with OI, where tooth extraction may be risky (risk of jaw fracture or alveolar socket fracture), and preventive and conservative treatment methods should be applied as much as possible. In patients with OI; Since poor oral hygiene can lead to caries and periodontal problems, oral hygiene standards must be met.

In this regard, it is extremely important to inform and educate parents and children by specialist dentists. In addition, the oral conditions of the erupted teeth should be followed and preventive and protective applications should be made when necessary.

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