

Osteogenesis Imperfecta with Dentinogenesis Imperfecta – A case report

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I. Introduction

Osteogenesis imperfecta (OI) is a serious inherited disorder which is commonly of an autosomal dominant pattern, however autosomal recessive and non hereditary types are also known to occur. The clinical features commonly observed in patients with osteogenesis imperfecta include blue sclerae, deficiency of growth such as short stature, bone fragility causing multiple fractures, hearing loss, hypermobility, joint laxity and dentinogenesis imperfecta.^[1-3] The disease causes either the production of structurally defective collagen or a decrease in collagen synthesis, hence all tissues rich in type I collagen may be affected.^[1,4] Four types of OI exist based on the classification of Silience et al^[5] [Table 1]. Peterson and Wetzel have evaluated the recent findings in classification of osteogenesis imperfecta by means of existing dental symptoms^[6] [Table 2].

II. Case Report

A five year old child reported to clinic and parents gave history of trivial fall following which he had pain and swelling over the left forearm and patient gave history of multiple fractures in lower limb within a span of 2 years. There was no relevant family history.

On examination, the child was found to be very fragile, with short stature and thin built and blue sclerae. X ray of the forearm revealed transverse fracture of ulna X ray chest showed barrel chest and X ray of long bones showed bowing with old fracture of femur and bowing of both bones of leg

Patient complained of pain in his tooth since the last 3 months following which dental examination was done which revealed a history of pain in the lower right and left posterior teeth. Intraoral examination showed caries in deciduous maxillary and mandibular right and left deciduous first and second molars. Discoloration of the primary teeth was evident. An OPG showing severe bone loss in relation to mandibular first molars was evident.

The above mentioned history along with clinical and radiological findings confirmed the diagnosis as Osteogenesis imperfecta type I with dentinogenesis imperfecta type I.

III. Discussion and review of literature

OI is a rare disorder. It has variously been called “osteopsathyrosis idiopathica” by Lobstein, “fragilitus osseum”, hereditary fragility of bone, later brittle bone disease and fragile bone disease, but the current name “OI” was coined by Vrolik in 1845.^[7,8] The disorder was first brought to the limelight by the Swede, Olof Jakob Ekman in his doctorate thesis at Uppsala in 1788 as “osteomalacia congenita” involving three generations.^[9,10] Since then, various researchers have tried to describe and classify this disorder into various types: In 1906, Looser, proposed the classification: “Congenita” and “tarda” based on time of fractures.^[11]

The most widely acceptable classification was by Silience *et al*

Table 1: Clinical types of osteogenesis imperfecta (Silience et al.)⁵

Osteogenesis imperfecta – type I Osteogenesis imperfecta tarda Osteogenesis imperfecta with blue sclera

Osteogenesis imperfecta – type II Osteogenesis imperfecta congenita (neonatal, lethal) Vrolik type of osteogenesis imperfecta

Osteogenesis imperfecta – type III Progressively deforming, normal sclerae

Osteogenesis imperfecta – type IV Osteogenesis imperfecta with normal sclera

Dental abnormalities

DI is a localized mesodermal dysplasia affecting both primary and permanent dentition. DI is an inherited disorder affecting dentin. Mutation in dentin sialophosphoprotein (DSPP) is the cause for this defect. The DSPP

gene is located at 4q21.3 in a cluster of dentin and bone matrix genes. DSPP encodes both dentin sialoprotein (DSP) and dentin phosphoprotein (DPP) as one precursor protein that is cleaved before secretion. DSP and DPP have different roles in dentinogenesis. DPP serves as a nucleator of mineralization and induces apatite formation.⁽¹²⁾

Histopathology

The dentin is composed of irregular tubules, often with large areas of uncalcified matrix. The tubules tend to be larger in diameter and less numerous in a given volume of dentin than in normal teeth.⁽¹³⁾

Table 2: Genetically conditioned dysplasias of dentin⁽⁶⁾

| | |
|----------------------------------|--|
| Dentinogenesis imperfecta type 1 | Displays manifestations of osteogenesis imperfecta |
| Dentinogenesis imperfecta type 2 | No manifestations of osteogenesis imperfecta |
| Dentinogenesis imperfecta type 3 | Brandywine type showing shell teeth |
| Dentin dysplasia type 1 | Radicular dentin dysplasia |
| Dentin dysplasia type 2 | Coronal dentin dysplasia |

The disease is usually inherited as an Autosomal Dominant (AD) trait although recently mutations are also reported in some cases.⁽¹⁴⁾ Patient had no family history of OI, so it was assumed that it is an AD trait or a new genetic mutation. However, because of the distinctive signs and symptoms, the early diagnosis of OI was made so there was no need significant finding. Features were indicative of OI type 1. All patients suffering from OI display which was similarly seen in our case. The aim of the treatment included strengthening of bone by reducing the rate of fractures as well as to prevent deformities of long bone using bisphosphonates and by giving appropriate splints.

Dentiogenesis imperfecta (DI) has been noted in 80% of OI types I, III and IV⁽¹⁵⁾ According to DI classifications this patient was assigned to DI type I, as the other two types of DI merely demonstrate dental signs and do not accompany with any type of OI.⁽¹⁵⁾ Intraoral examination revealed discoloration of the primary teeth whereas permanent dentition remained unaffected . Some areas of enamel fractures and attrition were also noted. Other dental anomalies such as ectopic eruption and missing (10%) are more common in OI⁽¹⁶⁾ There was no missing or ectopic eruption in our case. However, at the time of screening the patient was too young to detect any missed eruption due to his age. The patient was advised for prophylactic dental treatment.



Figure 1 - Caries in deciduous maxillary and mandibular right and left deciduous first molars



Figure 2 - 5 yrs old boy with osteogenesis imperfecta with frontal bossing



Figure 3 – Barrel chest



Figure 4- transverse fracture of ulna



FIGURE 5 – Bowing of Right Femur



Figure 6 – Bowing of both bones (left lower limb)

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