



Osteogenesis Imperfecta/Lobstein Syndrome associated with Dentinogenesis Imperfecta

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ABSTRACT

Osteogenesis imperfecta is a collagen related disorder characterized by increased bone fragility and low bone mass. The important oral finding in osteogenesis imperfecta is the presence of dentinogenesis imperfecta. This article presents a case of osteogenesis imperfecta (type IV B) with dentinogenesis imperfecta where a 7-year-old girl had opalescent primary teeth associated with severe bone deformity, scoliosis, barrel shaped rib cage, and short stature. The clinical, radiographic and histologic features are reviewed along with management aspects.

Keywords: Osteogenesis imperfecta, Dentinogenesis imperfecta, Bisphosphonate therapy.

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INTRODUCTION

Osteogenesis imperfecta (OI) is a heritable systemic disorder of connective tissue particularly affecting the tissues in which the principal matrix protein is type I collagen (mainly bone, dentin, sclerae and ligaments). Dentinogenesis imperfecta (DI) is a possible associated symptom of OI and belongs to the group of genetically conditioned dentin dysplasias. This disorder is caused by mutations in the genes (COLA 1 and COLA 2) that encode the pro- α 1 and pro- α 2 polypeptide chains of type I collagen.¹ Molecular genetic studies of OI have identified many independent mutations in both chains of type I collagen which leads to quantitatively or qualitatively deficient fibrils.² These mutations can be specific to a family or to an individual when they occur de novo. This condition had been referred by various synonyms over the years which includes Lobstein syndrome, Vrolik syndrome, brittle bone disease, glass bone disease, Porak and Durante's disease.³

This disorder has been recognized long back with studies beginning in 1788 with Swede OI of Jakob Ekman. The name OI dates back to at least 1895 and has been the usual medical term in the 20th century to present. The most widely used and accepted classification is given by Silence et al in 1979 based upon disease severity and progression. Presently with the application of modern bone histology techniques further subdivisions of the disorder with seven subtypes has been defined.²

CASE REPORT

A 7-year-old girl was referred to Department of Oral Medicine and Radiology with a chief complaint of brittle discolored teeth since eruption (Fig. 1). Both parents gave no previous history of this type condition on either sides of family. Her prenatal and natal history was unremarkable. Her medical history suggested primary consultation with pediatricians and orthopedicians for fracture of her legs twice (Fig. 2). Radiographical evaluation revealed treatment for the same.

On general examination patient stature appeared to be short with spinal curvature and barrel shaped rib cage. Bowed upper and lower limbs was also evident and the



Fig. 1: Dental status and opalescent tooth appearance



Fig. 2: Fractured right femur



Fig. 4: Bowing of tibia and fibula

patient was nonambulatory (Fig. 3). Clinical oral examination revealed all erupted discolored deciduous teeth which was opalescent brownish color with smooth polished dentin surface. Upper central and lateral incisors were extensively destructed and showed grade II mobility. Radiograph confirmed bowed upper and lower limbs (Fig. 4). On the basis of clinical radiographical features and opinions from medical specialists a diagnosis of OI type B was made. Patient was advised stainless steel crowns to 64, 74 and 84; glass ionomer restorations with canines and primary mandibular second molars, 54 was restored with amalgam restoration. Extraction of primary upper central incisors was done as it was grossly destroyed. Patient is being recalled every 2 months for review.

DISCUSSION

Osteogenesis imperfecta is an autosomal dominant disorder of connective tissue caused by the mutation in the genes



Fig. 3: Clinical skeletal abnormality

COL1A1 and COL1A2 that encode pro- α 1 and pro- α 2 chains of type 1 collagen which is the major protein of dentin and bone.¹ Most of the mutations that cause OI type 1 occur in COL1A1 gene. These mutations lead to reduction in the quantity of structurally normal type 1 collagen in the body. The mutations in case of type II, III and IV can occur in COL1A1 or COL1A2 gene. These mutations result in quantitative and qualitative alterations in the collagen synthesis. Mutations in CRTAP gene may be responsible for rare cases of OI.

Osteogenesis imperfecta is considered to be an orphan disease because of low incidence.³ The varying range of incidence has been reported from 1:1000 to 1:2000 births. Besides bone fragility it can result in features like laxity of ligaments, blue sclera, growth retardation, scoliosis and dentin dysplasia.⁴ Our case exhibited clinical feature of sillage type IV osteogenesis imperfect characterized by brittle bones, scoliosis, short stature, pathologic fractures and DI. Dentinogenesis imperfecta is one of the most important oral finding of this disorder which is characterized by presence of opalescent yellowish brown colored brittle teeth and can affect both primary and secondary dentition. Enamel may be of normal thickness but gets dislodged easily because of smooth dentinoenamel junction exposing the softer dentin. Radiographically pulp chamber and root canals may be partially or totally obliterated and may also present shortened roots and bulbous crown with constriction at the cervix of the crown. Histologically the dentin is characterized by embedded cells, interglobular dentin, irregular and reduced dentinal tubules.⁵

Regarding the dental management of children the treatment is challenging and should be attempted as early as possible with a multidisciplinary approach. Strict oral hygiene instructions and preventive treatment is important.

The aims of dental treatment for children with DI associated with OI are to ensure favorable conditions for eruption of the permanent teeth and normal growth of the facial bones and temporomandibular joint. The treatment of DI is focused on protecting the affected dentin from caries, attrition, abrasion and erosion. The options for restorative treatment usually include crowns. The rapid attrition of such teeth in patients with DI results very soon in a closed bite. The crowns should be reconstructed on deciduous and permanent molars as soon as they appear into the oral cavity, while even short delays result in wearing of the enamel crown to the gingival line.

Although there is no absolute systemic treatment for patients affected with this condition treatment should be aimed at increasing overall bone strength to prevent or reduce bone fracture and maintain mobility.⁶ Physiotherapy to strengthen muscles and physical aids to support can be used. The medical treatment based on biphosphonate (pamidronate)^{6,7} has been gaining importance from long time for the treatment of OI. Our patient is also on intravenous pamidronate therapy for every 4 months for 2 years. Biphosphonates improves the bone strength by increasing the quantity of bone, but it does not improve the quality of the bone as the gene defect is still present.^{8,9} Some studies show that growth hormone⁶ might be useful in combination with biphosphonates. Currently cell and gene based therapies is in the early stages of preclinical research.³ May be in the future genetic therapy might be an answer for this pathology.

CONCLUSION

It is very important to recognize this orphan disease as early as possible as it needs to be differentiated from child abuse. Also these patients need to be reviewed quite regularly to improve their lifestyle. Our patient is being recalled every 2 months to review the erupting permanent dentition and also advised to continue the drug pamidronate to increase the bone density/mass and also review the hearing as there might be hearing loss with age.

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