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# Dentinogenesis Imperfecta associated with Type 1 Osteogenesis Imperfecta: A Case Report

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## CASE REPORT

A 14-year-old girl presented to the department of oral medicine and radiology with a complaint of yellowish brown discolouration of teeth since childhood. She also complained of rapid wearing away of front teeth and increased sensitivity to cold. There was similar discolouration of her milk teeth as informed by her parents. She had experienced multiple lower limb fractures since birth which was evaluvated and diagnosed with Osteogenesis Imperfecta at 3 months of age. She was under regular follow up by an orthopaedician and for the past one year there was no history of fractures. She was born out of a non consaguinous marriage in a normal uneventful pregnancy. She has two sibilings and none of them are affected with similar condition.

On physical examination the higher mental functions were normal. She was short statured with significant facial asymmetry. There was bluish discolouration of sclera. Bilateral bowing of lower limbs were present. She showed class 3 skeletal pattern with midfacial hypoplasia and concave facial profile.

**Abstract:** Dentinogenesis Imperfecta is an autosomal dominant disorder of dentine affecting primary and permanent dentition. Mutation in dentin sialophospho protein on the long arm of chromosome 4 results in this defective dentin. The clinical appearance of teeth is characteristic showing an amber like translucency and severe attrition. It can also occur in association with osteogenesis Imperfecta, a genetic disorder of type 1 collagen. Here we present a case of Dentinogenesis Imperfecta associated with Type 1 Osteogenesis Imperfecta in a 14-year -old girl. **Keywords:** Dentinogenesis Imperfecta, Osteogenesis Imperfecta.

## INTRODUCTION

Dentinogenesis Imperfecta (DI) is an inheritable disorder of tooth development that results in the structural defects of dentin. It is an autosomal dominant trait affecting both primary and permanent dentition. It can occur as an isolated defect due to the mutation of dentin sialophosphoprotein gene on chromosome 4 as well as in association with osteogenesis imperfecta in 50% of cases [1]. Osteogenesis Imperfecta (OI) is a heterogeneous group of genetic disorders of type 1 collagen, primarily characterised by osteopenia and increased bone fractures. Most of the OI results from mutations in the genes (COL1A1 and COL1A2) that encode the pro alpha 1 and pro alpha 2 polypeptide chains of type I collagen [2]. Various other oral manifestations such as midfacial hypoplasia, posterior crossbite, class 3 malocclusion are also reported in OI. Here we report a case of DI associated with mild form of OI in a 14-year -old girl.

Intraoral examination revealed generalized opalescent teeth with greyish purple and brownish discolouration. Generalised chipping of enamel was present. Moderate amount of supragingival calculus with marginal gingivitis was noted. Localised attrition of anterior teeth, anterior cross bite between 12 and 42, posterior cross bite of right side, dental midline shift of 1.5mm to the right side were noticed.

Panoramic radiograph revealed generalised involvement of all the teeth with bulbous crowns and marked cervical constriction with obliteration of pulpal chambers.

Standing plain radiograph of both lower limbs showed lateral bowing of bilateral tibia and fibula with deformity of distal femur. Bones appeared osteoporotic. Epiphysis visualised normally.

Based on clinical and radiographic examination diagnosis of Dentinogenesis Imperfecta associated with Type 1 Osteogenesis Imperfecta was made.



Fig-1: Opalescent appearance of anterior teeth with heavy calculus deposits and gingival inflammation.



Fig-2: Amber like translucency of posterior teeth with rapid wear of 36.



Fig-3: Posterior crossbite on right side

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Fig-4: Significant facial asymmetry



Fig-5: blue sclera of eyes.



Fig-6: Bulbous crowns and marked cervical constriction with obliteration of pulpal chambers on panoramic radiograph.



Fig-7: Standing plain radiograph of lower limbs showed lateral bowing of bilateral tibia and fibu la with deformity of distal femur



Fig-8: short stature of the girl.

#### DISCUSSION

Dentinogenesis Imperfecta is a localised mesodermal dysplasia affecting dentine. It is an autosomal dominant disorder involving permanent and deciduous dentition. Mutation in dentin sialophospho protein gene on the long arm of chromosome 4 results in this defect. This gene encodes for dentin sialoprotein and dentine phosphoprotein, both having a significant role in dentinogenesis. DI was first classified by shields in 1973 into three main types.

- Type I associated with Osteogenesis Imperfecta (OI)
- Type II not associated with OI; also known as Hereditary opalescent dentin
- Type III-DI of the "Brandywine type" which was found in the Brandywine triracial isolate in Southern Maryland [3]. This classification system was well accepted but extensive studies have proven DI and OI are separate and discrete entities, requiring a need for revised classification.

A revised classification was proposed in 1999 where DI is classified as Type 1 and 2. Both types are not associated with OI. DI1 corresponds to DI Type II and DI2 to DI Type III of Shields classification, respectively. There is no substitute for shields Type I DI in this revised classification [4].

The clinical appearance of teeth is characteristic showing an amber like translucency with a color ranging from yellowish to bluish grey. Enamel will rapidly wear off from underlying abnormal dentine when subjected to occlusal stress. Schwartz and Tsipouras suggested that attrition of permanent teeth was less severe than in the primary teeth [5]. Hodge *et al.*, in 1940 observed that the progression of carious lesions in affected teeth is slow, mainly due to the rapid attrition of defective dentine [6].

Radiographically, teeth may resemble tulip shape with broad crowns and marked cervical constriction. Pulpal obliteration is an another remarkable finding. In this reported case, the clinical and radiographic findings were typical of Dentinogenesis Imperfecta.

Osteogenesis Imperfecta (OI) which is also known as brittle bone disease is a heterogeneous group

of genetic disorders of type 1 collagen, primarily characterised by osteopenia and increased bone fractures. Secondary features reported are short stature, blue sclerae, dentinogenesis imperfecta and hearing loss. Most of the OI results from mutations in the genes (COL1A1 and COL1A2) that encode the pro alpha 1 and pro alpha 2 polypeptide chains of type I collagen. Over 500 mutations in COL1A1 and COL1A2 have been reported. The incidence of OI is 1 per 20,000– 30,000 live births [7].

In 1979, Sillence et al., classified OI into basic Types I–IV. Type I is an autosomal dominant mild form of OI. Patients present at preschool age with blue sclera and hearing deficits in 50% of cases. Type II is an autosomal recessive form, in which patients present with blue sclera and perinatal death. Type III is considered to be the most severe form of the disease, is an autosomal recessive form characterized by a progressive short stature and blue sclera that normalize with age. Type IV is an autosomal dominant form with moderate severity. Patients present with normal sclera and hearing; bowing bones and vertebral fractures are also common findings [8]. Most cases (90%) are classified as Type I or IV, with or without involvement of the teeth. The case reported here is thought to be Type 1 OI which is the mild form inherited as autosomal dominant trait. The patient had presented with history of recurrent fractures of lower limb, blue sclera, dentinal defects but with no hearing deficit. Craniofacial abnormalities were also observed in this case. Jensen et al., found that the abnormal posture, weight and size of the head in the OI population, might lead to the development of malocclusion [9]. The moresevere abnormalities of craniofacial features were seen with the more-severe types of OI in adult patients. O'Connell et al., reported that class III dental malocclusion occurred in 70% to 80% of types III and IV of the OI population, with a high incidence of anterior and posterior crossbites and open bites [10]. The case reported here also showed skeletal class 3 pattern with midfacial hypoplasia and posterior crossbite.

Silence classification have been expanded in 2004 with addition of type 5, 6, 7 OI. Later in 2007, OI type VIII was also added to this [11]. These types of OI are not associated with type 1 collagen mutations but presents with a similar phenotype and microscopically abnormal bone.

Abnormal dentin architecture in OI result from mutations in the gene COL1A1 and COL1A2 with a resultant qualitative and quantitative defect of type 1 collagen. Ultrastructural findings in dentin have suggested that a functional network consisting of matrix molecules collagen 1, fibronectin, tenascin and growth factors will be involved in terminal differentiation of odontoblast. Secondary to abnormal metabolism of type 1 collagen, the migration, terminal differentiation,

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secretion and life cycle of odntoblast will be abnormal [12]. Thus abnormal dentinogenesis in OI could be explained on the basis of odontoblast dysfunction. Other Syndromes associated with dentinogenesis imperfecta are Ehlers Danlos syndrome, Goldblatt syndrome, Schimke immunoosseousdysplasia and Brachioskeletogenital syndrome [13].

The early dental treatment is recommended for children with DI to ensure favourable conditions for eruption of the permanent teeth and normal growth of the facial bones and temporomandibular joint. Treatment includes caries prevention, observation of regressive changes of the teeth, monitoring the development of the craniofacial skeleton, and placement of artificial crowns. Both mixed and permanent dentition demands for multidisciplinary approach and it is often challenging.

#### CONCLUSION

Dentinogenesis Imperfecta can present as a secondary feature in type 1 collagen disorders such as Osteogenesis Imperfecta. A case of DI associated with OI will be characterised by fragile bones and dentinal defects. Early comprehensive dental treatment should be recommended in such cases for a favourable aesthetics, occlusion and functioning of teeth.

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