International Journal of Research in Medical Sciences
Gopal SK et al. Int J Res Med Sci. 2023 Feb;11(2):735-740
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012
DOI: https://dx.doi.org/10.18203/2320-6012.ijrms20230199

Case Report

Dentinogenesis imperfecta type 1-Shield's classification (1973): a rare case report and literature review

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Received: 25 December 2022
Accepted: 12 January 2023

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ABSTRACT

Dentinogenesis imperfecta (DI) is an autosomal dominant disorder that affects the dentin development. It is characterized by the presence of opalescent dentin, with dusky blue to brownish discoloration of the teeth. This condition is genetically and clinically heterogeneous; DI may affect only the teeth or it may be associated with the osteogenesis imperfect (OI). When both are associated there are various degrees of clinical presentation from mild form with less significant signs to severe form such as death in infancy. The most familiar classification system of this hereditary disorder is Shields classification which is of 3 types namely, type I, DI associated with OI, type II is not associated with OI. Type III which is also known as 'Brandywine type' DI. This case report describes a rare case of 32-year-old female patient who showed the characteristic features of Shield's classification type I with penetrance of defect through 4 generations.

Keywords: Dentinogenesis imperfecta, Osteogenesis imperfecta, Opalescent dentin, Shield's classification, Developmental defect, Dentin

INTRODUCTION

Developmental defects in structure of teeth includes enamel defect, dentin defects and cementum defects. Dentinogenesis imperfecta (DI) is a hereditary developmental dentin defect that results in the opalescent teeth. It is a localized mesodermal dysplasia affecting both the primary and permanent dentitions.[1]

The disease is inherited in an autosomal dominant fashion with high penetrance and a low mutation rate.[2,3] It was probably first recognized by Barret in 1882.[4] Robert and Schour in 1939 coined the term, 'DI'.[5]

DI has a reported incidence of 1:8000 whites in the United States. In a cross-sectional study done in North America, approximately 50% of osteogenesis imperfecta (OI) children present with dental abnormalities in various degrees and DI is the most prominent oral characteristic in these patients.[6] Gupta et al in 2011, did a cross sectional study in Indian population and found prevalence and distribution of DI as 0.09% which is among the rarest percentage of developmental dental anomalies.[7] There are various classification systems of this DI as shown in Table 1.

The most familiar classification system of this hereditary disorder is that formulated by Shields in 1973 which is of 3 types namely type I, DI associated with OI, type II, DI not associated with OI (hereditary opalescent dentin) and type III, 'Brandywine type' DI. Witkop in 1975 named the 3 types as dentinogenesis imperfecta, hereditary opalescent dentin, and brandywine isolate.[8,10]

A revised classification was proposed where DI was classified into 2 types with no substitute for DI associated with osteogenesis imperfecta (Table 1).[11,12]

Table 1: Classification of DI.

Shields et al[8]	Witkop et al [10]	Revised[11, 12]	Clinical presentation
DI-type 1	DI	No substitute	OI with opalescent teeth
DI-type II	Hereditary opalescent teeth	DI- type I	Isolated opalescent teeth
DI-type III	Brandywine isolate-found only in a population of southern Maryland (USA)	DI- type II	Isolated opalescent teeth

CASE REPORT

A 34-year-old female patient, reported to the department of oral medicine and radiology in Meenakshi Ammal Dental College and Hospital with the chief complaint of yellowish stains in her teeth since childhood. Patients also complaints of pain in her right lower back tooth region past 1 month which is dull and intermittent. Patient gives history of yellow discolouration of her primary and permanent teeth since childhood with frequent chipping of upper and lower teeth while chewing. On eliciting past medical history patient gives history of fracture of left elbow when she was 9 years old and it healed by itself. Her personal history reveals consanguineous marriage with son of paternal aunt and has a 12-year-old son. Patient had another son who died at the age of 3 due to unknown cardiac defect 2 years ago. Family history reveals her paternal grandmother, father and paternal aunty and uncle, her brother and her son were affected similar discolouration of teeth. Her brother has hearing defect since child hood. Another brother and her own son died at the age of 2-3 years due to unknown cardiac defect. Figure 1 explains the pedigree chart of our patient.

On general examination patient has short neck and is not able to extend her left hand completely with exuberant callus formation at the elbow of her left hand. On extra oral examination eyes appear white sclera with mild blue tinted hue near the cornea of both the eyes evident (Figure 2). Mandibular hypoplasia evident. Intraoral examination reveals presence of bluish grey opalescence of upper and lower teeth with chipping of enamel noted in 17, 31, 32, 41, 45, 47. (Figure 3). Grossly decayed 47 with tender on percussion. Correlating patient's history, family history, clinical features provisional diagnosis DI type I (Sheild's classification) was given.

Further patient was subjected to intra oral peri apical radiograph in relation to 41, 31, 32 (Figure 4) region which reveals partial loss of coronal tooth structure involving enamel, dentin and pulp with reduced radiopacity and obliteration of root canal space up to apex with intact lamina dura evident. Orthopantomogram (Figure 5) was taken which revealed presence of bulbous coronal portion with prominent cervical constriction and blunt roots noted which gives tulip shaped teeth with partial obliteration of root canal up to middle third of the roots in molars and premolars evident. Also, missing 18, 28, 36, 37, 46, grossly decayed 48, dental caries 16 and 26, prominent antigonial notch, hypoplastic and flattening of condyle and shallow sigmoid notch was evident. Lateral cephalogram revealed hypoplastic mandible and lordosis of neck spine (Figure 6). Lateral skull view and posteroanterior (PA) view of skull reveals presence of numerous Wormian bones noted in coronal and lambdoid suture (Figure 7) and X-ray of left elbow reveals presence of exuberant callus formation on the lateral epicondyle of left humerus bone (Figure 8). Patient was also subjected blood investigation which reveals normal serum calcium and phosphorus and mild increase in alkaline phosphatase (297 IU/l). Patient underwent extraction of grossly decayed 48 which was given for histopathological investigation of ground section which reveals irregular bundled dentinal tubules in crown and root portion and obliteration of root canal space. Correlating the history, radiographic features and histological features final diagnosis of DI type I (Sheild's classification) was given. Patient was managed with full mouth rehabilitation using prosthetic crowns.

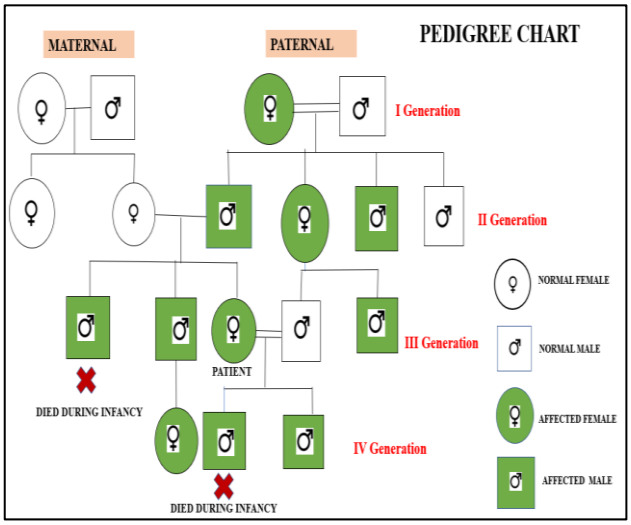

Figure 1: Pedigree chart of the patients.

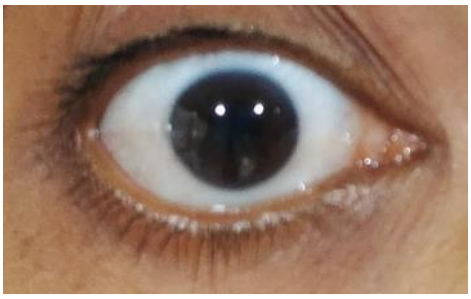

Figure 2: Eyes appears white sclera with mild blue tinted hue near the cornea of both the eyes evident.

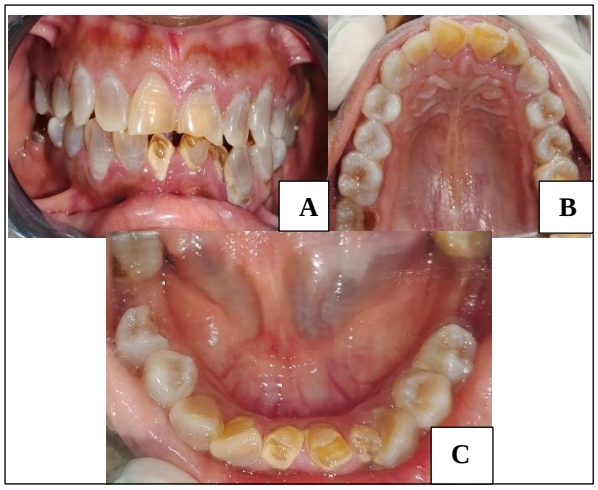

Figure 3: (A) Bluish grey opalescence of teeth; (B) maxillary arch; and (C) mandibular arch.

Figure 4: IOPA 41, 31, 32.

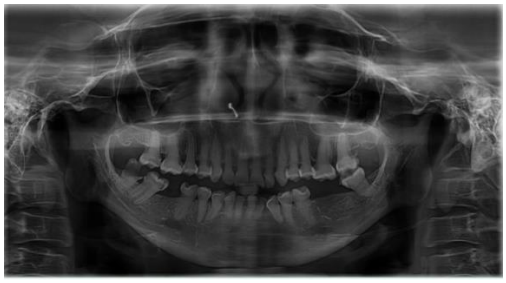

Figure 5: OPG showing tulip shaped teeth.

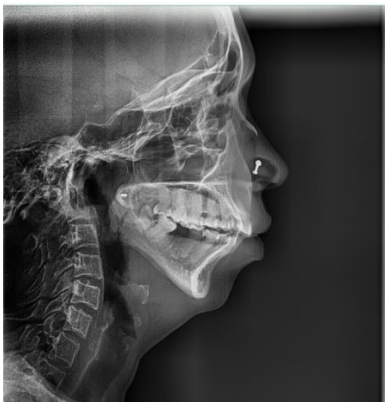

Figure 6: Lateral cephalogram showing hypoplastic mandibel and lordosis of neck.

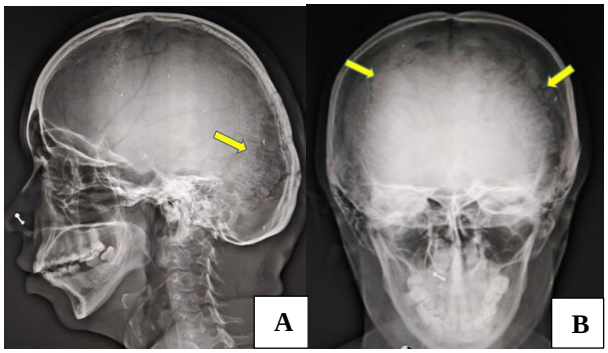

Figure 7: (A) Lateral skull view; and (B) PA view sowing wormian bone in coronal and lambdoid sutures.

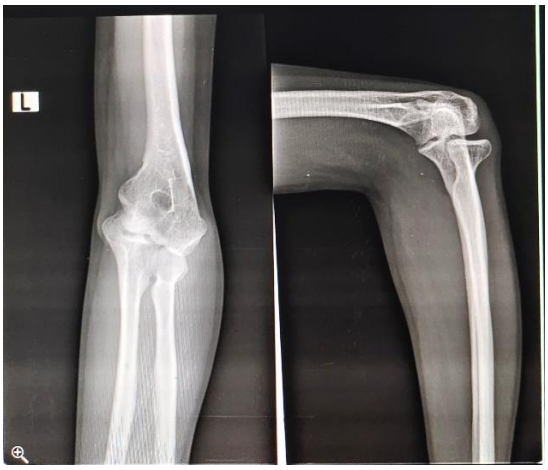

Figure 8: Anteroposterior and lateral view of maximum extended left arm shows exuberant callus formation on the lateral epicondyle of left humerus bone.

DISCUSION

Dentin is the mineralized tissue that forms the bulk of the crown and root portion of tooth. Dentin is rigid but elastic tissue which contains large numbers of small and parallel tubules in a mineralized collagen matrix. Dentin consists of 65% inorganic material, 35% organic matter and water. The inorganic component consists of hydroxyapatite crystal units [3Ca3 (PO4)2 · Ca (OH)2], as in bone, cementum, and enamel. The organic substance consists of collagenous fibrils embedded in the ground substance of mucopolysaccharides (proteoglycans and glycosaminoglycans). Type I collagen is mainly found in the dentin. The important constituents of the ground substance are the proteoglycan, glycoproteins like dentin sialoprotein, osteonectin, osteopontin; phosphoproteins like dentin phosphoprotein, gamma carboxyglutamate containing proteins and phospholipids. The protein of dentin matrix and bone are similar, but dentin sialoprotein and dentin phosphoprotein are present exclusively in dentin, which helps in mineralizing the dentin. They also contain certain growth factors like fibroblast growth factor, tissue growth factor etc.[13,14]

DI is localized form of mesodermal dysplasia which occurs during histodifferentiation stage. It is a gene defect in structural or the regulatory protein namely dentin sialo phosphoprotein (DSPP) which is mapped in location chromosome 4q21.3. This precursor protein is cleaved into 2 dentin specific proteins namely, dentin sialoprotein and dentin phosphoprotein.[15-19] This leads to under mineralized dentin and obliterates the pulp cambers and canals giving appearance of opalescent teeth. Examination of family pedigree in our patient revealed that an affected child had an affected parent. Poornima et al reported DI I in 3 generations, where as we have reported DI I involving four generations.[20] Thus, this indicates the complete penetrance of the defective gene in the family.

Clinically, the appearance of the teeth with DI is characteristically showing a high degree of amber like translucency and colour ranging from yellowish brown to bluish grey which gives an opalescent sheen, when observed under transmitted or reflected light. The enamel may be hypoplastic or hypocalcified in about one-third of the patients and tends to crack away from the defective dentin. Severe and rapid attrition exposes dentin.[21] Affected teeth have broad crowns with constriction of the cervical area which gives the teeth a tulip shape. Dental tissues in DI will have low hardness, elasticity and stiffness leading to a phenomenon of micro movement resulting in failure of restorations.[22] In adults, they may frequently wear down at gingival region and stains the exposed dentin to dark brown or even black. Some patients demonstrate an anterior open bite.[23] All these clinical findings were in accordance with present case.

Radiographically, the teeth appears solid, lacking pulp chambers and root canals, slight to marked attrition of the occlusal surface, short and slender roots and constriction at the cervical portion of the teeth giving the crown a bulbous appearance. Types I and II DI usually shows partial or complete obliteration of the pulp chambers which was evident in our case. In the early stage of development of teeth, large pulp chambers may be evident, which quickly obliterates by the formation of dentin which leads to absence of root canals or thread like.[23,24] Similar findings were observed in our case.

Histologically, the enamel, although normal in structure, tends to crack. The DEJ is not scalloped but appears flattened although it appears qualitatively normal. The structure of the mantle dentin may also be normal, whereas the dentinal tubules in circumferential dentin appears coarse, branched and with reduced number of tubules. The presence of an atubular area in the dentin with reduced mineralization and a reduced number of odontoblasts are consistent findings. Pulpal inclusions, increased interglobular dentin and large areas of unmineralized dentin with irregular borders between the unmineralized and mineralized dentin are seen. Odontoblasts entrapment within the dentinal matrix may also be evident.[25,26] Syndromes associated with DI are OI, Ehlers Danlos syndrome, Goldblatt syndrome, Schimke immune osseous dysplasia, Brachioskeletogenital syndrome.[27] Among the syndromes associated, the common one is OI which is also called as brittle bone disease. It is a heritable connective tissue disorder with an incidence of approximately 1 in 10000 births. Males and females are equally affected and it can occur in all racial and ethnic groups. It comprises a heterogeneous group of heritable disorders characterized by impairment of collagen maturation heterozygosity for mutations in one of two genes that guide the formation of type I collagen namely the COL1A1 gene on chromosome 17 and the COL1A2 gene on chromosome 7. Collagen I form a major portion of bone, dentin, sclerae, ligaments, and skin.[28] Clinical classification based on disease severity given by Sillence et al in 1979 is a continuum, varying widely from prenatal lethality, extreme bone fragility to nearly asymptomatic without bone fracture.[29] Clinical manifestations are blue sclera, sometimes mild protrusion, hyper extensibility of joints or contractures, hearing defect, DI, easy bruising, variable height, spine defects like scoliosis, kyphosis, fracture that healed by itself with abnormal callus formation. Craniofacial findings include triangular facies, frontal bossing, macrocephaly flattened vertex and skull base, prominent occiput, maxillary or mandibular hyperplasia or hypoplasia, malocclusion crossbite, open bite, impacted first and second molar, chin sharply pointed, softening of bone and flattening of sides of the mandible.

Radiographic appearance shows multiple fracture, bowing, angulation or deformity of the long bones, skeletal deformity, osteopenia and wormian bones in skull suture. Histology of bone shows immature spongy bone, while the trabeculae of the cancellous bone are delicate and often show microfractures. Serum concentration of vitamin D, calcium, phosphorus are normal but alkaline phosphatase may be normal or slightly elevated.[28,30,31] In our case patient had similar history of hearing defect and children who died at an early age due to cardiac defects, and clinically mild blue sclera with mild proptosis, skeletal deformity with exuberant callus, lordosis, hypoplastic mandible and wormian bones evident in skull radiographs and serum alkaline phosphatase was slightly elevated. Hence our case was diagnosed as dentinogenetic imperfecta with features of osteogenesis imperfecta according to Shield's classification type I.

The management of DI must be done with considerations on the fact that the entire dentition is at risk because of numerous problems. The root canals become threadlike and may develop micro exposures, resulting in periapical inflammatory lesions. There is always a risk of enamel loss and significant attrition, they are not good candidates for full crowns because of the chances of cervical fracture.

The success of full coverage is best in teeth with crowns and roots that exhibit close to a normal shape and size. Overlay dentures covered with fluoride-releasing glass ionomer cement have been successfully used in some cases. For OI the fractures may be a major problem which should be managed with physiotherapy, rehabilitation, and

orthopedic surgery. Medical treatment with intravenous or oral bisphosphonates can provide clinical benefits, including decreased pain, reduced risk of fractures, and improved mobility- reserved for moderately to severely affected patients.[31]

CONCLUSION

Eliciting patient's medical history and family history is very important in diagnosis of DI, because it can be a manifestation associated with syndromes like OI. Radiology and histology are important investigations in their diagnosis. An efficient full mouth rehabilitation was done aiming at establishing a more favourable prognosis for such a complex anomaly. Hence appropriate treatment must be advised along with preventive measures, to achieve a good aesthetic result and restore function.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Gopal SK, Poongodi V, Priyadharshini S. Dentinogenesis imperfecta type 1-Shield's classification (1973): a rare case report and literature review. Int J Res Med Sci 2023;11:735-40.