Case report

Dental findings in patient with brittle bone disease

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Abstract:

Osteogenesis imperfecta (OI) or brittle bone disease is genetically heterogeneous connective tissue disorder which is characterized by skeletal deformities due to fragile bone, reduced bone mass and frequent fractures. The genetic mutations in collagen genes, COL1A1 and COL1A2 are responsible for the pathogenesis of OI. The clinical and radiological features of OI manifest in different age groups and severity of the condition depends on the type of OI. The common clinical findings include recurrent and multiple fractures, laxity of the ligaments, blue sclera, growth retardation, and scoliosis. OI is commonly associated with Dentinogenesis imperfect; (DI) which is characterised by defective dentin formation. Here we report a

rare case of OI type IV (Group A) with dental manifestations such as micrognathia, retained primary teeth, class III malocclusion, crossbite, multiple impacted teeth, and delayed eruptions.

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Introduction: Osteogenesis imperfecta (OI) is a hereditary congenital osteoporosis, characterized by bone fragility and low bone mass, and is linked to a mutation in the gene encoding type-I collagen. It exhibits a variety of clinical presentation ranging from intrauterine death to normal growth and low fracture incidence depending upon the severity. The incidence of OI varies between 6 and 20 in 100,000 new-borns and its prevalence is 4-10 in 100,000 individuals.[1]Sillence classified, OI based on clinical, genetically, and radiographic features into four groups.

Type I is a mild form and Type II is the lethal form. Type III patients show progressive limb deformation. Patients with type IV shows moderate to severe phenotypes and do not fit into any of the first three categories. [2]Some cases of OI demonstrate heterogeneous features, which are not mentioned in Sillence classification. Patients affected by these types do not demonstrate DI and blue sclera;OI types V-VIII are called syndromes resembling OI. [3,4] Sillence's type IV is further subdivided into groups A and B, which can be differentiated based on the dental findings. Group B is characterized by the presence of Dentinogenesis imperfect (DI) and group A is associated with normal teeth. [5]

Dentinogenesis imperfecta (DI) is a hereditary disorder of dentin. The mutation of gene dentin sialophosphoprotein result in the formation of defective dentin that is prone for wear and fracture. Shields et al proposed three types of DI: type 1 is associated with OI. Type 2 has essentially the same clinical radiographic and histological features as DI type 1 but without OI; Type 3 is rare and is only found in the triracial Brandywine population of

Maryland.[6]The most common clinical manifestations in teeth are teeth discoloration and enamel fracture. The enamel may be of normal thickness, but frequently is dislodged exposing the softer dentin which may be attributed to the smooth dentinoenamel junction. We report a case of type IV Group A OI with normal enamel and dentin.

Case History

A 20-year-old female patient reported to SEGi oral health centre with the complaint of pain in left lower jaw region. Pain was mild, intermittent and aggravates during chewing. Her past medical history includes multiple fractures in long bones (femur and pelvic) since childhood and these fractures were treated by specialists. Both the parents and siblings did not have any bony diseases. On general physical examination, she had short stature (Figure 1), normal upper and lower limbs, normal gait, size of the head appeared smaller however, it was in proportionate to her body stature.

Figure 1: Photograph showing short stature

On extraoral examination, she had normal mandibular movements and adequate mouth opening. Intraoral examination showed micrognathia of both the arches, mixed dentition with retained 53,55,63,64,65, 73,74,75 and 85 (Figure 1 &2).

Mild brownish discoloration of deciduous teeth with conical shaped 23 and missing 14,15, 16, 17, 23, 24, 25, 26, 27 33, 34, 35, 37, 45 were noticed.Class III dental malocclusion, cross bite and mild to moderate attrition of retained teeth was evident.

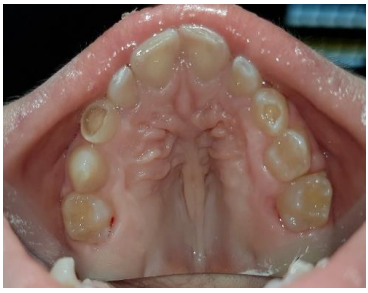

Figure 2: Photograph of maxillary arch

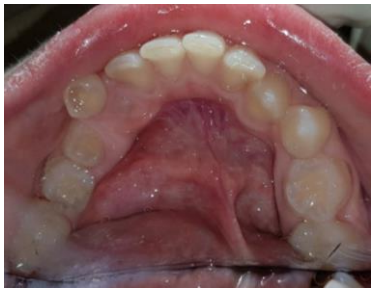

Figure 3: Photograph of mandibular arch

OPG revealed normal thickness and enamel and dentin. Size of the pulp chambers appeared normal. Multiple impacted 14,15, 16, 17 23, 24,25, 26,27, 34, 35, 45 and 47 were noticed. There were structural and morphological changes in right and left condylar head without any evidence of fractures (Figure 4).

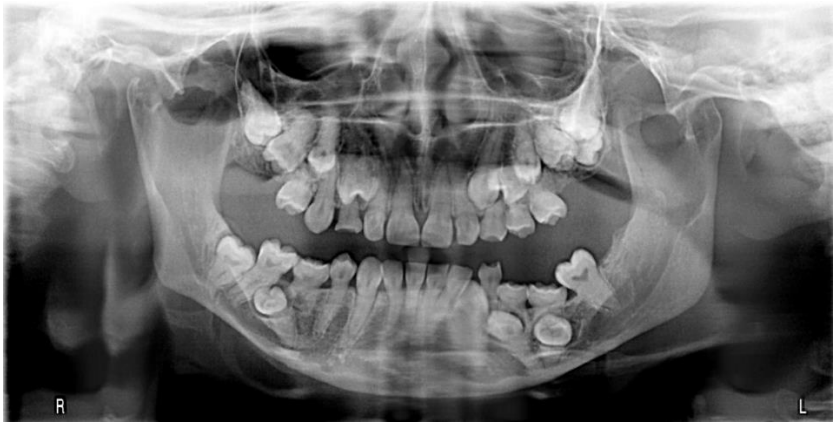

Figure 4: OPG showing multiple reatined primary teeth and impacted permanent teeth.

Based upon these clinical findings and previous medical history, a provisional diagnosis of osteogenesis imperfect (Type IV A) with normal dentin was made. Patient was explained regarding the dental condition and condyles. The preventive dental care was rendered and informed to come for periodic follow up.

Discussion:

Osteogenesis imperfecta, also known as "brittle bone disease", is a heterogeneous group of genetic connective tissue-associated disorders.[7]Most form of OI is due to mutations in the genes (COL1A1 and COL1A2) that encode the pro-alpha 1 and pro-alpha 2 polypeptide chains of type I collagen. Hence, tissues which have abundant Type I collagen like bone, dentin, sclera, and ligaments are usually affected most.[7]A reduced amount of structurally normal collagen results in OI Type I, whereas qualitative and quantitative alterations in the collagen synthesis

result in OI Types II, III, and IV.[8]Three possible reasons that may cause a child to be born with OI. [9]

1. Direct inheritance from a parent: overall there are 50% chance that the disorder will be passed to next generation.

2. A new dominant mutation: spontaneous gene mutation of the sperm or the egg before the child's conception.

3. Mosaicism: clinically an unaffected parent may have more than one affected child. In this scenario mutation may have occurred during fetal development of the parent.

In our case, patient's clinical feature showed moderate form of OI, Sillence Type IV, which is characterized by brittle bones, growth retardation, pathologic fractures, without DI. Such moderate form has better survival rate compared to severe form which occurs at a very young age. [10]Sillence's Type IV OI is the most diverse group as it contains all OI cases which cannot be categorized in Type I to Type III. Type IV is subdivided into A and B based on their dental findings.[9]Type IVA is associated with normal teeth whereas Type IV B has DI Type II and teeth may

appear as opalescent greyish brown hue. Even though enamel of teeth in Type IV B will be of normal thickness, it easily gets dislodged due to soft dentin and smooth dentinoenamel junction. Ideally, diagnosis of OI should be achieved at earliest to provide adequate dental treatment to reduce the need for extensive and invasive procedures.[7] Sometime diagnosis of OI can be challenging, especially if the family history is negative. In such scenario clinical examination and genetic counselling can aid in diagnosis. [11]

Severe types of OI has abnormal craniofacial characteristics due to various malocclusion. In one report, class III malocclusions occurred in 70–80% of types III and IV OI cases, with a high incidence of anterior and posterior cross bites and open bites. [12] In our case, patient had class III malocclusion with cross bite. Her micrognathic jaws led to dental findings of impacted permanent teeth and retained deciduous teeth with no changes in enamel and dentin thickness.

The medical treatment of OI is focused on minimizing fractures, maximising mobility, independent function and general health.

Pamidronate, a type of drug which increases bone density and regulates bone formation has thus far shown considerable success in the treatment of severe OI. Safe exercises such as swimming are encouraged to promote a healthy lifestyle. Orthopaedic surgery to implant rods may be recommended to increase support to bones.[10]

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