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Case Report / Olgu Sunumu

Dental Findings and Treatment in Osteogenesis İmperfecta: A Case Report

Osteogenezis İmperfektada Dental Bulgular ve Tedavi: Bir Vaka Raporu

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ABSTRACT

Osteogenesis imperfecta (OI) is a hereditary disorder characterized by heightened bone fragility, reduced bone density, and connective tissue abnormalities. The condition, commonly referred to as 'glass bone disease', is characterized by various symptoms, including the blue sclera, hearing impairments, deformities in the hands and feet, as well as dentinogenesis imperfecta. This case presents a 10-year-old female with Osteogenesis Imperfecta (OI) who submitted an application to the Department of Pedodontics at Gaziantep University Faculty of Dentistry. Among the physical findings of the patient, short stature, hearing loss, pectus excavatum, grayish blue sclera, hand-foot deformities, and malocclusions in the relationship between the jaws were found. In the clinical examination of the patient, lack of oral hygiene, deep dentin caries, crowding, macroglossia, class 3 malocclusion were observed, and teeth that needed treatment were treated.

Keywords: Osteogenesis imperfecta, Pedodontics, Dental findings

ÖZ

Osteogenezis imperfekta (OI), artmış kemik kırılganlığı, düşük kemik kütlesi ve bağ dokusunda bozuklukların görüldüğü genetik geçişli bir hastalıktır. Diğer bir deyişle 'cam kemik hastalığı'; mavi sklera, işitme problemleri, el-ayak deformiteleri ve dentinogenezis imperfekta ile ilişkilendirilmiştir. Bu olguda, Gaziantep Üniversitesi Diş Hekimliği Fakültesi Pedodonti Anabilim Dalı'na başvuran 10 yaşında Oİ tanılı kız çocuk hasta sunulmaktadır. Hastanın fiziki bulguları arasında boy kısalığı, işitme kaybı, pectus ekskavatum, grimsi mavi sklera, el-ayak deformiteleri, çeneler arası ilişkide maloklüzyonlar saptanmıştır. Hastanın klinik muayenesinde oral hijyen eksikliği, derin dentin çürükleri, çapraşıklık, makroglossi, sınıf 3 maloklüzyon görülmüş ve tedavi gereksinimi olan dişleri tedavi edilmiştir.

Anahtar Kelimeler: Osteogenezis imperfekta, Pedodonti, Dental bulgular

Introduction

Osteogenesis imperfecta (OI) is a prevalent hereditary condition characterized by bone fragility. It encompasses a diverse range of genetic disorders primarily attributed to defects related to type 1 collagen. The condition is transmitted in an autosomal dominant manner in approximately 85-90% of instances and is attributed to mutations occurring in the COL1A1, and COL1A2 genes, resulting in deficiencies in type 1 collagen, either quantitatively or quatitatively. In the past decade, there have been reports on the identification of abnormalities in additional proteins that participate in the regular processing of type 1 collagen. The collagen that forms the extracellular structure of tendon, skin, and bone tissues is Type 1 collagen. Therefore, osteogenesis imperfecta patients have problems in these tissues.[1] In this disease, increased bone fragility is caused by mineralization defects and deterioration of the bone matrix. To standardize the management of OI cases, a multidisciplinary approach by orthopedists, physiotherapists, dentists, and other allied health professionals is required.[2]

Developed in 1979, the Silence classification categorized patients with OI into four subtypes ranging from mild to mortal according to clinical severity. OI is divided into four main types, depending on the severity of bone fragility and clinical/radiologic features:

Type I OI, mild bone deformity, OD inherited; Type II OI, blue sclera, no progressive deformity, prenatally or perinatally lethal, OR inherited; Type III OI, severe form, OR inherited, progressive bone deformity, characteristic face, the most severe type associated with perinatal survival. Type IV OI is a less well-defined form of moderate severity (phenotype between types I and III).[3] The Silence classification remains useful, but as new gene defects have been identified, different classifications have been created.[4,5]

From a clinical perspective, symptoms of OI exhibit heterogeneity and demonstrate a range of severity. In addition to skeletal symptoms, this condition has the potential to impact various systems, such as dental and craniofacial structures, muscle strength, auditory function, and respiratory and cardiovascular health. Advisable to adopt a multi-disciplinary approach to provide care and treatment that encompasses not only fractures, diminished mobility, growth, and bone pain but also various non-skeletal symptoms. Although bisphosphonates continue to be the primary treatment for OI, researchers are investigating alternative approaches, such as sclerostin inhibitor antibodies and TGF beta inhibition, to target both low bone density and the underlying bone fragility.[6]

Craniofacial and dental symptoms also vary according to the type of disease. There may be triangular facial structure, severe skull growth disorder, malocclusions, dentinogenesis imperfecta (DI), oligodontia, or unerupted teeth. From a clinical perspective, teeth that are impacted by DI exhibit a range of discoloration, which can vary from gray-brown to blue in appearance. Several radiographic findings have been associated with the condition, including altered root morphology, bulb-like crowns, pulp obliteration, taurodontism, and a high prevalence of dental caries. DI is frequently observed in individuals diagnosed with OI, with a prevalence that varies within the range of 8%. It is worth noting that not all teeth are necessarily impacted by OI. For instance, it is commonly observed that primary teeth tend to exhibit a greater degree of discoloration in comparison to permanent teeth. The permanent first molars have a higher tendency to experience discoloration compared to the anterior teeth. It is important to note that not all unaffected teeth may show signs of pulpal obliteration or taurodontism.[3,7]

Furthermore, alongside OI certain individuals exhibit DI, a hereditary dentin anomaly distinguished by the presence of teeth with a grayish-blue to brown discoloration and pulp obliteration. Currently the field of DI is classified into three distinct sub-types. DI type I commonly linked with OI and arises due to genetic mutations occurring in the COL1A1 and COL1A2 genes, which are responsible for encoding collagen type I. DI types II and III are attributed to genetic mutations occurring in the gene responsible for encoding dentin sialophosphoprotein (DSPP). It is important to note that these specific types of DI are not observed in patients diagnosed with OI. In the context of DI, it is observed that the enamel exhibits a typical structural appearance, yet it lacks proper support from the underlying atypical dentin. Dentin is predominantly comprised of hydroxyapatite, an organic phase consisting of type 1 collagen and water, in terms of its structural composition. Defects in dentin arise as a result of mutations occurring in the genes responsible

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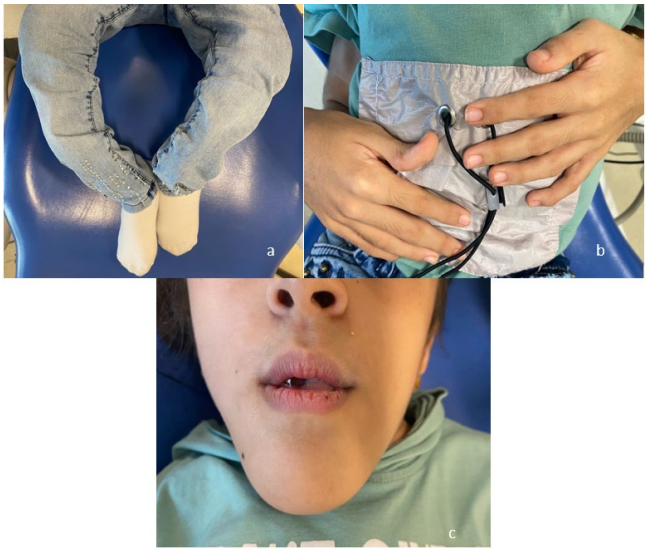
for encoding type I collagen. In individuals with DI type I, both primary and permanent are commonly impacted, with the primary dentition typically exhibiting more severe manifestations compared to the permanent dentition.[8,9]

The objective of this case report is to present a comprehensive analysis of the dental treatment and orofacial manifestations observed in a patient with OI.

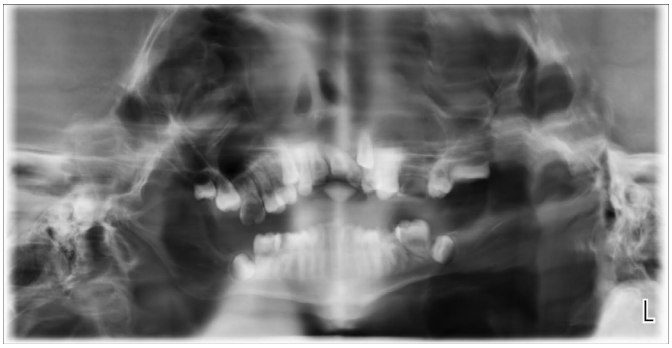
Case report

In January 2023, an 11-year-old girl was admitted to Gaziantep University, Faculty of Dentistry, Department of Pedodontics with the complaint of pain and inability to feed. All procedures to be performed, possible complications, and the use of photographs for scientific purposes were explained in detail to the parents and the child, and an informed consent form and child consent form was obtained. A consultation was requested from Gaziantep University Faculty of Medicine Hospital, Department of Child Health and Diseases regarding the patient's risk status in terms of dental treatments. It was informed to us that there is no risk other than performing the procedure without neck extension and protecting the airway. In her medical history, multiple operations for bone fractures and the use of Zoledronate were reported. At the same time, it was reported that her 5-year-old brother was also diagnosed with osteogenesis imperfecta. Although the parents did not have a diagnosis of OI, the father was reported to have a skeletal deformity in the lower extremity.



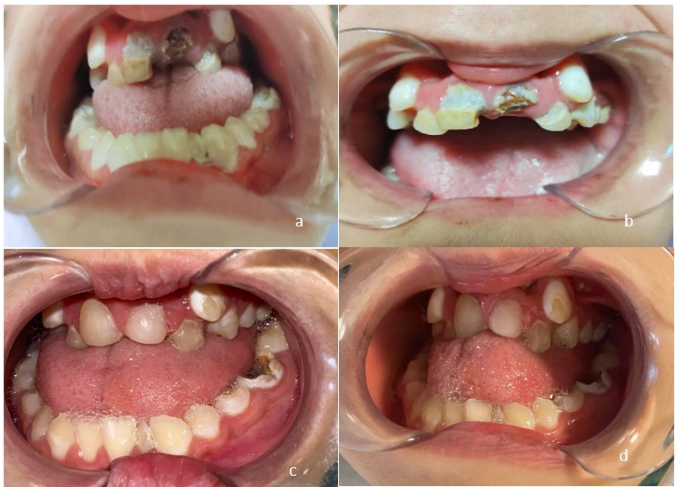
Picture 1. Extraoral findings (a:foot, b:hand, c:jaw)

Extraoral examination revealed graying of the whites of the eyes, pectus excavatum, severe scoliosis, short stature, hand and foot deformities, short stature, weakness, and inability to walk and swallow (**Picture 1**). Her parents reported that she was fed only liquid formula. Only orthopantomography x-ray (OPG) (Morita veraview IC5 HD, Kyoto, Japan) (**Picture 2**) and limited mouth-opening photographs could be taken.



Picture 2. OPG image of the patient

Our patient had maxillary growth retardation, open bite, macroglossia, Class 3 malocclusion, crowding in the lower anterior region, reverse overjet, and labial proclination in the upper canine teeth. There were no problems with tooth color, but there were white lesions, deep dentinal caries, shortness of some tooth roots, and enlargement of the pulp chamber similar to taurodontism. In light of these symptoms, we think that the patient has type III OI. The OPG radiography of the patient at the first session showed that there was no missing tooth (**Picture 2**). The patient's level of cooperation was evaluated as a score of 3 according to the Frankl scale.[10] Deep dentin caries and chronic apical periodontitis were found in teeth numbered 16-26-36-21. Cold and electric pulp tests were performed on these teeth and no sign of vitality was obtained. Deep dentin caries were found in teeth numbered 11-12-14-22-24-46. In teeth 13-23, cavitation was found in tooth 23 with white lesions. Composite restoration (G-aenial Composite□, GC, Tokyo, Japan) was applied to teeth 14-23-24 in the patient's first session. Fissure sealant (Fissurit FX, VOCO GmbH, Cuxhaven, Lower Saxony, Germany) was applied to teeth 15-34-35-44-45. In the 2nd session, coronal amputation with mineral trioxide aggregate (Angelus MTA®, Industria de Produtos Odontologicos Ltda, Londrina, Brazil) was performed on teeth 11-12-22-46. They were restored with composite filling (G-aenial Composite®, GC, Tokyo, Japan) using a resin-modified glass ionomer cement (GC Fuji II LC Capsule®, Tokyo, Japan) base. In the 3rd session, tooth number 21 was treated with root canal treatment and restored with a strip crown (Tdv Dental, Brasil). In the 4-5th session, teeth numbered 16-26-36 were indicated for extraction. In the patient's consultation response, it was stated that treatment should be performed by preventing neck hyperextension. It has been reported that treatment under general anesthesia is not appropriate due to the narrow respiratory tract and the drawback of hyperextension. Moreover; It has been reported that broad-spectrum antibiotic treatment is necessary to minimize the risk of secondary infection and osteonecrosis. Extractions were left to the last sessions due to the risk of possible medication-related osteonecrosis of the jaws (MRONJ) and infection, and primary closure was performed after extraction. As the patient required tooth extraction and could not be postponed, the patient was consulted with a medical doctor, and bisphosphonate use was suspended for 1 month before extraction and 3 months after extraction. The patient was called for a follow-up visit for tissue healing and was told that medication should not be started. A broad-spectrum antibiotic treatment was started 5 days before tooth extraction, primary closure was provided after extraction, and antibiotic treatment was continued for 20 days. CHX mouthwash was recommended when the patient had problems with spitting, but the parents were able to apply topical application to the extraction site with cotton wool. Low-dose laser therapy (biostimulation) is applied during and 1 week after the extraction. It was applied 3 times (Sirolaser advance plus, Dentsply Sirona, USA) for 1 minute. This treatment has been suggested to accelerate tissue regeneration, reduce bacterial colonization in the area and reduce the risk of MRONJ.[11]



Picture 3. a,b: Untreated image / c,d: Treated image

Parents were told about tongue and mechanical teeth cleaning 3 times a day after each feeding. Parents were advised to give the child a drink of water after the use of sugar-containing formula. The use of an electric toothbrush was recommended to her parents due to her limited mouth opening. Regular dental check-ups every 3 months were recommended since the patient was in the high caries risk group (**Picture 3**). Fluoride varnishes are intended to prolong the contact time of fluoride with the enamel surface. Varnishes usually contain 5% NaF (22000 ppm F-). It is used for its advantages such as being easy to administer and accepted by young children, reducing the risk of fluoride ingestion.[12] Because of swallowing dysfunction in our patient, fluor varnish (Imicryl®, Polimo, Turkey) was applied instead of fluorine gel and CCP-ACP derivatives (Tooth Mousse™, GC, Switzerland) were recommended twice a day after brushing. The combined use of casein phosphopeptide amorphous calcium phosphate (CPP-ACP) and fluoride varnish has been reported to provide a greater reduction in lesion depth than when used alone. The use of CPP-ACP-containing products is recommended after brushing with fluoride toothpaste, especially in high-risk children. The use of these products is successful in the remineralization of white spot lesions.[12,13]

Discussion

Osteogenesis imperfecta (OI), is an uncommon hereditary skeletal condition by variable levels of bone fragility. It manifests in approximately 1 in 10,000 to 20,000 infants at birth.[14] OI patients need to be rehabilitated with a multidisciplinary approach due to serious systemic problems. The treatment process progresses with medication, orthopedic treatment, physical therapy, dental treatment, and psychological support. Patients are often faced with bone fractures and pain, so traumatic, major surgeries are not preferred in dental treatment.

Given that patients frequently utilize bisphosphonate-derived medications (BP), it is imperative to incorporate medical consultation as an integral component of the treatment procedure. These patients benefit from the utilization of drugs that possess an antiresorptive effect. Surgical dental procedures in patients undergoing bisphosphonate therapy have been identified as a potential risk factor for the development of osteonecrosis of the jaws, commonly referred to as medication-related osteonecrosis of the jaw (MRONJ). Frequent occurrence of eruption disorders of permanent teeth is observed in pediatric patients with OI. Nevertheless, there is a lack of established protocols for the extraction of deciduous teeth in pediatric patients with OI who are undergoing bisphosphonate treatment. Previous studies have documented that around 40% of individuals diagnosed with OI who were administered bisphosphonates experienced deciduous tooth extractions as a result of eruption disorders, with no observed complications. The bone phenotype of OI can contribute to the complexity of DI. Regarding the OI type, it is advisable for dentists to consult the information provided by the medical doctor and develop personalized dental treatment plans accordingly.[15]

Novel pharmacologic treatments currently under investigation include teriparatide[16] in adult OI and denosumab[17] in pediatric patients. Both interventions have demonstrated favorable outcomes in recent clinical trials. The development of mesenchymal stem cell therapy for OI is currently underway, and initial experiments conducted on mouse models have demonstrated encouraging outcomes.[18]

DI is the main oral problem associated with OI. Osteogenesis imperfecta patients, especially dentinogenesis imperfecta patients, are recommended for periodic oral health management. Stainless Steel Crowns and occlusion elevation were recommended for teeth affected by DI after restoration.[19] In OI patients, dentinogenesis findings may not be seen as in our patients. In our case, discoloration and oligodontia were not observed, but bulb-like enlargements in the pulp chamber and onion-like crowns were seen in the posterior region. All these symptoms were also detected in the 5-year-old brother of our patient in our clinic.

It was reported in the literature that a number of caries, periodontal and orthodontic problems are common in these patients.[20] Our patient had a high number of caries and caries depth with gingivitis. Therefore, family oral hygiene education, additional oral care advice, and regular professional medical support were recommended.

OI patients often require orthodontic problems and orthognathic surgery. Orthodontic treatment may have limitations due to bone structure defects and weakness, depending on the type of disease and the systemic condition of the patient.21

Conclusion

In patients diagnosed with OI, medical, social, and dental anamnesis is very important. Their medications, social life, diet, and oral hygiene habits must be known. Treatment and oral care of special patients, including skeletal conditions that affect dentition, must be carried out in cooperation with parents. Conservative treatment of all dental treatment of patients diagnosed with OI should be conservative and should aim to restore as much function as possible. It is important for the identification and multidisciplinary management of patients in terms of regular specialist physician control and preventive-interceptive treatments.

Authors' Note

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It is declared that during the preparation process of this study, scientific and ethical principles were followed and all the studies benefited are stated in the bibliography.

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Yazar Katkıları / Author Contributions

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Veri Toplanması | Data Acquisition: OA (%90), YI (%10)
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Makalenin Yazımı | Writing up: OA (%70), YI (%30)
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REFERENCES

1. Tournis S, Dede AD. Osteogenesis imperfecta–a clinical update. Metabolism. 2018;80:27–37.
2. Ralston SH, Gaston MS. Management of osteogenesis imperfecta. Front Endocrinol (Lausanne). 2020;10:924.
3. Taqi D, Moussa H, Schwinghamer T, Ducret M, Dagdeviren D, Retrouvey J-M, et al. Osteogenesis imperfecta tooth level phenotype analysis: Cross-sectional study. Bone. 2021;147:115917.
4. Glorieux FH, Rauch F, Plotkin H, Ward L, Travers R, Roughley P, et al. Type V osteogenesis imperfecta: a new form of brittle bone disease. J Bone Miner Res. 2000;15(9):1650–8.
5. Semler O, Garbes L, Keupp K, Swan D, Zimmermann K, Becker J, et al. A mutation in the 5′-UTR of IFITM5 creates an in-frame start codon and causes autosomal-dominant osteogenesis imperfecta type V with hyperplastic callus. Am J Hum Genet. 2012;91(2):349–57.
6. Marom R, Rabenhorst BM, Morello R. Management of endocrine disease: Osteogenesis imperfecta: An update on clinical features and therapies. Eur J Endocrinol. 2020;183(4):R95–106.
7. O'connell AC, Marini JC. Evaluation of oral problems in an osteogenesis imperfecta population. Oral Surgery, Oral Med Oral Pathol Oral Radiol Endodontology. 1999;87(2):189–96.
8. Ibrahim S, Strange AP, Aguayo S, Shinawi A, Harith N, Mohamed-Ibrahim N, et al. Phenotypic properties of collagen in dentinogenesis imperfecta associated with osteogenesis imperfecta. Int J Nanomedicine. 2019;9423–35.
9. Nutchoey O, Intarak N, Theerapanon T, Thaweesapphithak S, Boonprakong L, Srijunbarl A, et al. Phenotypic features of dentinogenesis imperfecta associated with osteogenesis imperfecta and COL1A2 mutations. Oral Surg Oral Med Oral Pathol Oral Radiol. 2021;131(6):694–701.
10. Paglia L, Gallus S, de Giorgio S, Cianetti S, Lupatelli E, Lombardo G, et al. Reliability and validity of the Italian versions of the Children's Fear Survey Schedule - Dental Subscale and the Modified Child Dental Anxiety Scale. Eur J Paediatr Dent [Internet]. 2017 [cited 2021 Jun 15];18(4):305–12. Available from: https://pubmed.ncbi.nlm.nih.gov/29380617/
11. Günay AY, Gülsüm AK. İlaca Bağlı Olarak Çene Kemiklerinde Görülen Osteonekrozda Dental Yönetim ve Güncel Tedavi Yaklaşımları: Derleme. Sağlık Bilim İleri Araştırmalar Derg. 2020; 3(2):81–92.
12. Duraisamy V, Xavier A, Nayak UA, Reddy V, Rao AP. An in vitro evaluation of the demineralization inhibitory effect of F− varnish and casein phosphopeptide-amorphous calcium phosphate on enamel in young permanent teeth. J Pharm Bioallied Sci. 2015;7(Suppl 2):S513.
13. Kumar VLN, Itthagarun A, King NM. The effect of casein phosphopeptide-amorphous calcium phosphate on remineralization of artificial caries-like lesions: an in vitro study. Aust Dent J. 2008;53(1):34–40.
14. Forlino A, Marini JC. Osteogenesis imperfecta. Lancet. 2016;387(10028):1657–71.
15. Yoneda T, Hagino H, Sugimoto T, Ohta H, Takahashi S, Soen S, et al. Bisphosphonate-related osteonecrosis of the jaw: position paper from the allied task force committee of Japanese society for bone and mineral research, Japan osteoporosis society, Japanese society of periodontology, Japanese society for oral and maxillofacial radiology, and Japanese society of oral and maxillofacial surgeons. J Bone Miner Metab. 2010;28:365–83.
16. Orwoll ES, Shapiro J, Veith S, Wang Y, Lapidus J, Vanek C, et al. Evaluation of teriparatide treatment in adults with osteogenesis imperfecta. J Clin Invest. 2014;124(2):491–8.
17. Hoyer-Kuhn H, Franklin J, Allo G, Kron M, Netzer C, Eysel P, et al. Safety and efficacy of denosumab in children with osteogenesis imperfecta-a first prospective trial. J Musculoskelet Neuronal Interact. 2016;16(1):24.
18. Prockop DJ. The exciting prospects of new therapies with mesenchymal stromal cells. Cytotherapy. 2017;19(1):1–8.
19. Okawa R, Kubota T, Kitaoka T, Kokomoto K, Ozono K, Nakano K. Oral manifestations of Japanese patients with osteogenesis imperfecta. Pediatr Dent J. 2017;27(2):73–8.
20. Majorana A, Bardellini E, Brunelli PC, Lacaita M, Cazzolla AP, Favia G. Dentinogenesis imperfecta in children with osteogenesis imperfecta: a clinical and ultrastructural study. Int J Paediatr Dent. 2010;20(2):112–8.
21. Najirad M, Madathil SA, Rauch F, Sutton VR, Lee B, Retrouvey J-M, et al. Malocclusion traits and oral health–related quality of life in children with osteogenesis imperfecta: A cross-sectional study. J Am Dent Assoc. 2020;151(7):480–90.