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Case Report / Приказ болесника

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Oral manifestations and rehabilitation of a patient with *osteogenesis imperfecta*

Оралне манифестације и рехабилитација пацијента са *osteogenesis imperfecta*

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SUMMARY

Introduction Osteogenesis imperfecta is a rare heritable connective tissue disorder characterized by increased fragility of the bony tissue. The incidence of orofacial alterations associated with osteogenesis imperfecta is variable and includes dentinogenesis imperfecta, malocclusions, hypoplasia of the jaws, delayed dental development and structural abnormalities of the teeth.

Case outline A 22-year-old girl was referred to the Clinic for Pediatric and Preventive Dentistry for dental treatment. Enlarged head, triangular-shaped face, mandibular prognathism with excessive maxillary hypoplasia, lowered vertical occlusal dimension were present features. The intraoral findings included dentinogenesis imperfecta with Kennedy's class IV in the upper jaw and class II in the lower jaw. Panoramic radiograph revealed abnormalities in crown and root shape, obliteration of the pulp chamber and severe deficiency of alveolar bone mass. Overall treatment involved five phases: I – Preventive and prophylactic treatment, II – Direct restauration of five teeth with glass ionomer cement, III – Extraction of severely damaged teeth, IV – Prosthodontic rehabilitation with removable partial dentures, V – Maintenance and follow-up phase.

Conclusion Low prevalence and wide variety of signs and symptoms make dental treatment of osteogenesis imperfecta overly complex and challenging. Nevertheless, it is essential to improve craniofacial and dental function along with facial aesthetic.

Keywords: osteogenesis imperfecta; rare diseases; dentinogenesis imperfecta; partial dentures

САЖЕТАК

Увод *Osteogenesis imperfecta* представља ретко, наследно обољење везивног ткива које карактеришу крхке кости склоне фрактурама и прогресивни коштани деформитети. Неке од орофацијалних манифестација су *dentinogenesis imperfecta*, малоклузије, хипоплазија вилица, закаснели развој зуба и структурне аномалије зуба.

Приказ болесника Болесница је имала карактеристичне промене: увећану главу, троугласти облик лица, мандибуларни прогнатизам са израженом хипоплазијом горње вилице и сниженом вертикалном димензијом оклузије. Клиничким прегледом утврђена је *dentinogenesis imperfecta*, а на ортопантомограму уочене су аномалије облика крунице и корена зуба, облитерација пулпне коморе и недостатак алвеоларне коштане масе. Рехабилитација је обухватила неколико фаза: 1 – Превентивне и профилактичке мере; 2 – Рестаурација каријесних лезија; 3 – Екстракција зуба; 4 – Протетска рехабилитација; 5 – Контролни прегледи.

Закључак Стоматолошки третман овог комплексног ретког обољења захтева детаљну анализу и мултидисциплинарни приступ како би се постигла рехабилитација оралних функција, побољшала естетика лица, а тиме и квалитет живота пацијената.

Кључне речи: *osteogenesis imperfecta*; ретке болести; *dentinogenesis imperfecta*; парцијалне протезе

INTRODUCTION

Osteogenesis imperfecta (OI) is a rare heterogeneous group of connective tissue disorders characterized by increased fragility of the bony tissue.[1] Its estimated frequency in the general population is about 1 in 15.000 to 20.000 new-borns [2, 3]. Most patients have dominant mutations in one of two genes, *COL1A1* and *COL1A2,* which code the collagen type I synthesis [4, 5]. The genetic database has been updated with 18 gene mutations reported in a recent review [6]. These mutations lead to quantitative or qualitative changes in type I collagen, the

substantial structural protein of bone and dentin. Consequently, the quality of the osseus tissue is reduced and bones are more fragile and prone to fractures. These multiple fractures could follow minor trauma or sometimes occur spontaneously [7]. In addition to numerous fractures, patients might display short stature, hearing impairment, blue sclerae, skeletal deformities that affect craniofacial structures such as triangular facial form, large head size and soft calvaria [8]. The incidence of orofacial alterations associated with osteogenesis imperfecta is variable and includes dentinogenesis imperfecta (DI), maxillary hypoplasia, skeletal class III deformity, crossbite, open bite, hypodontia/oligodontia and delayed dental development [9-11]. Malocclusions can impair daily activities, such as chewing and speaking, which has negative impact on quality of life and serious psychological and social implications [12]. Also, affected teeth might have crowns with a bulbous structure, constriction in the cementoenamel junction, irregular root morphology, enlarged pulp chamber as well as pulp stones and obliterations [13, 14].

OI has diverse clinical expression, varying from very mild to severe with perinatal lethality [15]. Based on clinical findings, Sillence et al. described four types of OI and since than its classification has been continuously updated (Table 1) [16, 17].

To our knowledge, there are not many cases of prosthodontic rehabilitation of younger patients with OI reported in the literature. This rare disease is complex and requires multidisciplinary approach and medical expertise. The aim of this study was to present rare case of a patient with severe deforming type of Osteogenesis imperfecta and its dental treatment.

CASE REPORT

A 22-year-old girl was referred to the Clinic for Pediatric and Preventive Dentistry, School of Dental Medicine, University of Belgrade for dental treatment. She had been diagnosed with OI two days after birth. Postnatal skull radiography revealed constitutional bone fragility and decreased mineralisation and radiograph of the upper limbs showed left ulnar fracture with present mild angulation. Further examinations indicated that it was OI type III.

In the medical history she had multiple fractures of upper and lower extremities during childhood as well as skeletal deformities and bisphosphonate treatment. She had common craniofacial features including bluish sclerae, disproportionally enlarged head compared to the body, triangular-shaped face, spine deformity.

Clinical and radiographic examinations were performed to obtain a comprehensive evaluation of maxillomandibular complex. Mandibular prognathism with excessive maxillary hypoplasia was noted. The facial appearance was irregular with disproportionate inferior third and compression of the middle third. Consequently, vertical occlusal dimension was lowered.

The intraoral examination revealed dentinogenesis imperfecta. In the upper jaw, the frontal segment was edentulous and in posterior segments the existing teeth had caries lesions and direct restorations. In the lower jaw, lateral incisors, canines, left first premolar and right second molar were present and had brown opalescent hue. Furthermore, gingival recession was found in the buccal region of both lower lateral incisors and the rest of teeth showed excessive inclination of the crown and rotation. Posterior segments of alveolar mandibular ridge were underdeveloped and thin (Figure 1).

Panoramic radiograph was evaluated regarding abnormalities in crown and root shape, anomalies od pulp chamber and structure of the jaws. In both, maxilla and mandible, severe deficiency of alveolar bone mass was present. Affected teeth had bulbous shaped crowns with constricted cementoenamel junction, short roots, and complete obliteration of the pulp chamber (Figure 2).

Considering patient's medical history and complexity of the condition, a therapy was based on minimally invasive dental procedures with minor trauma. The expectations of the patient were analysed, and various treatment options were discussed. Overall treatment involved five phases: I – Preventive and prophylactic treatment, II – Direct restorations of teeth 14, 15, 17, 24, 25, 26 with glass ionomer cement, III – Extraction of teeth 23, 27, 47 in local anaesthesia, IV – Prosthodontic rehabilitation with removable partial dentures, V – Maintenance and follow-up phase.

Prosthodontic rehabilitation of the patient started after extraction wounds had healed. Preliminary impressions were made for both arches using irreversible hydrocolloid impression

material and study casts were obtained. Custom trays were made and used for definitive impressions. Occlusal rims were fabricated on the final casts and used to record maxillo-mandibular relationships. Adequate function, patient's facial characteristics and aesthetic and muscular tolerance were evaluated to determine the optimal vertical occlusal dimension. The waxed-up dentures were placed and evaluated in the mouth of the patient. After this phase, the dentures were finished, polished and after occlusal adjustment they were delivered to the patient (Figure 3, 4). Also, she was trained how to maintain proper oral and denture hygiene. An appointment was scheduled after a week for final adjustments and after that she was examined after 3, 6 and 12 months. She was satisfied with the functional improvement and with the aesthetic outcome of the treatment (Figure 5). The removable partial dentures did not need realignment after 1 year.

The present work was approved by competent ethics committee and conforms to the legal standards. Written informed consent for participation and publication, including clinical details and accompanying images, was obtained from the patient.

DISCUSSION

Osteogenesis imperfecta is a rare disease in which all parts of the body containing collagen type I can be affected, including skeletal system, dentin, dermis, tendons, organ capsules, fascia, meninges, cornea and sclera [18]. Literature data suggests that craniofacial and dental abnormalities are common findings [14, 19, 20]. The diagnostic procedure involves analysis of complete medical and family history, clinical examination and dental radiography. Previous studies suggested that multidisciplinary approach was needed to ensure accurate diagnosis and adequate treatment procedures [21]. Furthermore, dental team should include a paediatric dentist, a prosthodontist, a periodontist, an oral surgeon and an orthodontist.

Authors stated that OI type III was the most severe form in children which survive the neonatal period [16, 22]. These patients, along with OI type IV, require special dental care from the primary dentition [21]. Malocclusions are frequent finding in OI patients, especially class III [8, 23]. The malocclusions are caused by maxillary hypoplasia, mandibular prognathism or a combination of both factors. In addition, abnormal bone growth, posture, head size might be

contributing factors to the development of malocclusions, which may become more serious with time [20]. Malocclusions can impair everyday activities such as chewing and speaking and consequently have negative impact on quality of life [12, 24], which was one of the main concerns of our patient.

Another manifestation is dentinogenesis imperfecta which prevalence varies by OI type, from 21% to 73%, as reported in the literature [13]. Bulbous crowns, short roots, obliteration of the pulp chamber, as seen in our patient, can compromise some dental procedures [25].

Intravenous bisphosphonates (BPs) are the primary treatment of children with moderate to severe OI. The main mechanism of their action is inhibition of osteoclast function and bone resorption. The effect of BPs therapy on the dental tissues is still unclear [26]. One of the concerns is development of bisphosphonate related osteonecrosis of the jaws (BRONJ) following even simple teeth extractions [25]. Studies reported that no complications had been observed after extractions of the primary teeth in children with BPs treatment [21, 27]. In present case report, patient did not have any complications in the healing process after extraction of the permanent teeth.

Individuals with OI have disturbances in organic and mineral bone components and altered biomechanical characteristics resulting in brittleness of bones. Moreover, it is followed by insufficient amount of bone, the cortical thickness and decreased amount of trabecular bone [28]. Malmgren et al. reported that individuals with OI had a high prevalence of missing teeth, with a predilection for the posterior regions of the jaws [11]. Panoramic radiograph of our patient revealed underdeveloped upper and lower jaw. Additionally, she has multiple teeth missing and consequently unpreserved vertical occlusal dimension which made normal functioning, especially eating, exceedingly difficult. To establish physiological function, preserve alveolar bone and achieve acceptable facial aesthetic, we carefully planed rehabilitation of the orofacial system [1]. Severe type of OI, combined with potential complications and patients' rejection of orthognathic surgical procedure, based the therapeutic strategy on minimally invasive treatment. Due to our patients' lower financial status, it was determined that the therapy included partial dentures was the best option.

The dental management of OI patient primarily depends on medical history, patient's age and needs, social and economic circumstances. The low prevalence and wide variety of signs

and symptoms of OI, make the dental procedure complex and challenging. All present limitations and possible complications must be taken into consideration. However, the main goal is to improve craniofacial and dental function along with facial aesthetic. In this case report, functional and aesthetic rehabilitation was achieved, and the patient was successfully adapted to partial dentures.

Conflict of interest: None declared.

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Table 1. Classification of osteogenesis imperfecta

Type	Inheritance	Gene	Clinical feature
I	AD	COL1A1, COL1A2	Blues sclerae, normal stature, fractures, hearing loss, presence of DI rare
II	AD	COL1A1, COL1A2	Perinatal lethal, Blue-Gray sclera, small for gestational age, respiratory distress, limb deformities, "frog leg" positioning, soft calvarium
III	AD	COL1A1, COL1A2	Severe phenotype, short stature, multiple fractures, progressive deformities, may have DI, adolescent onset hearing loss
IV	AD	COL1A1, COL1A2	Milder than OI III, typically ambulatory, DI is common, adult-onset hearing loss, normal-gray sclerae
V	AD		Mild to moderate, calcification of the interosseus membrane, radial head dislocation, hyperplastic callous formation
VI	AR	SEFPINF1	DI absent, like type III
VII	AR	CRTAP	Overlap with types II and III, milder forms also documented
VIII	AR	LEPRE1	Overlap with types II and III, milder forms also documented

AD – autosomal dominant; DI – dentinogenesis imperfecta; AR – autosomal recessive; CRTAP – cartilage-associated protein

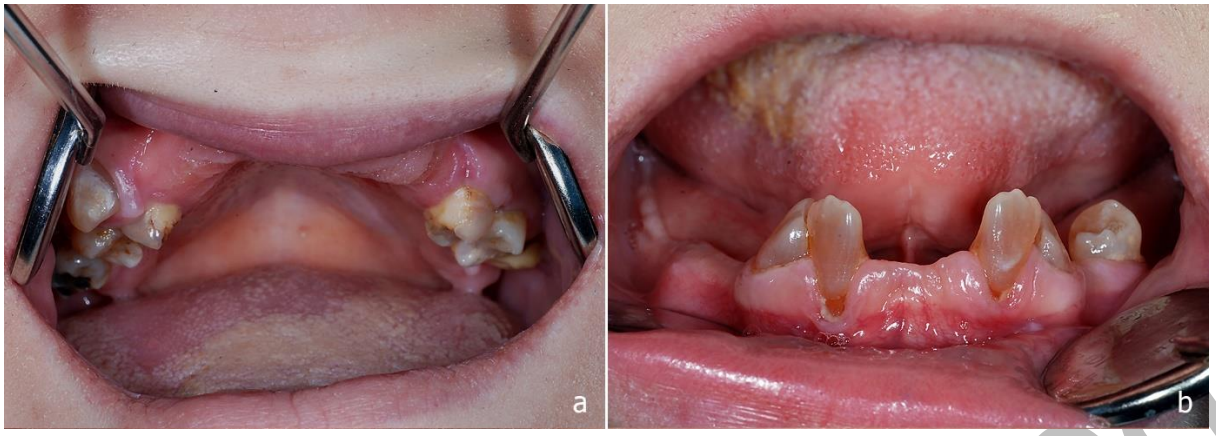

Figure 1 Clinical examination; intraoral photographs of the: a – maxillary, b – mandibular arch

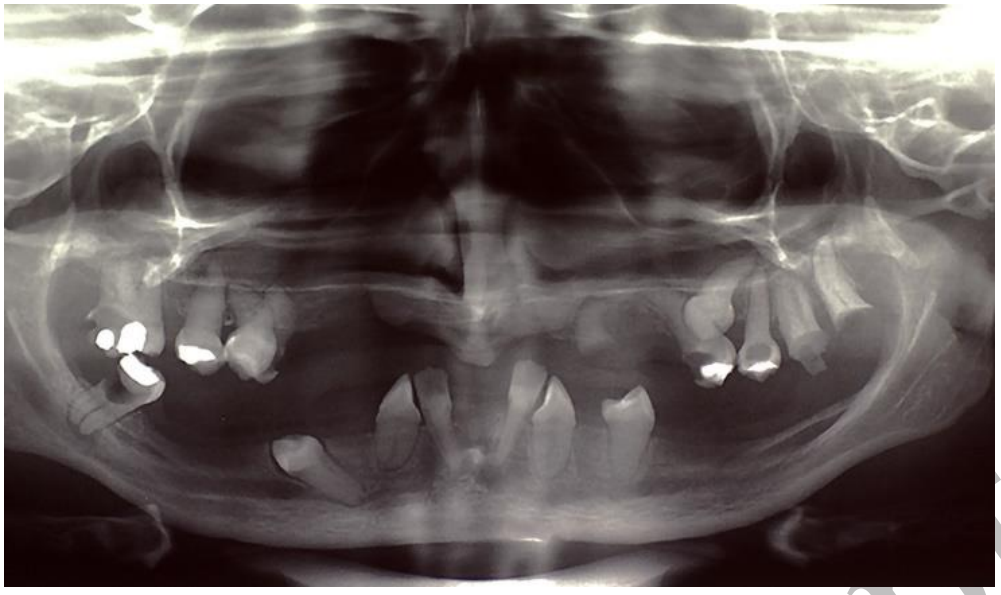

Figure 2. Radiographic evaluation: panoramic view showing irregular crown and root morphology of the present teeth; teeth with caries lesions and lower right second molar with periradicular radiolucency and a large loss of the crown; a residual root in region of the upper left lateral incisor; deficiency of alveolar bone mass in both jaws

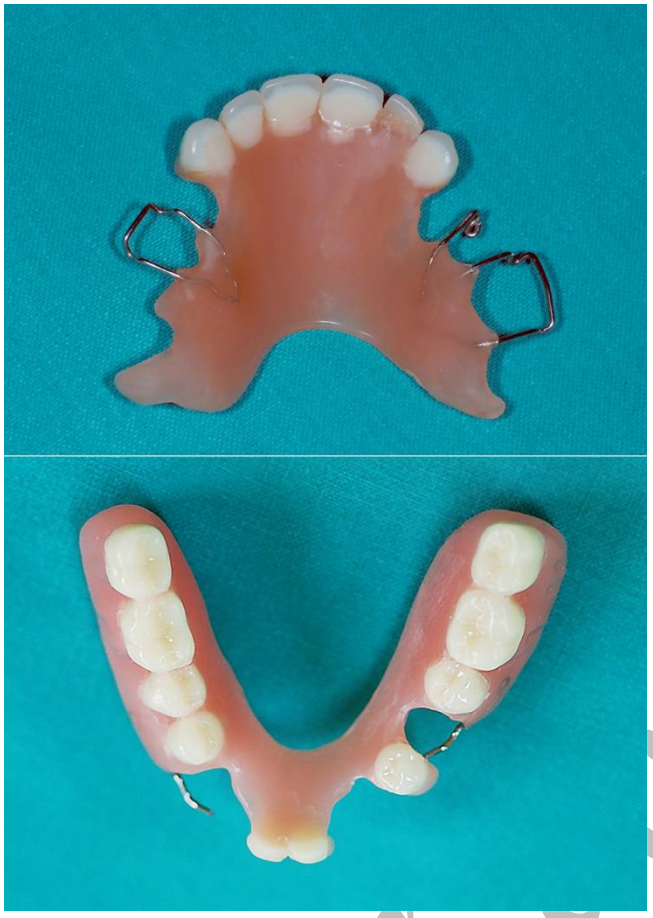

Figure 3. Maxillary and mandibular partial removable dentures.

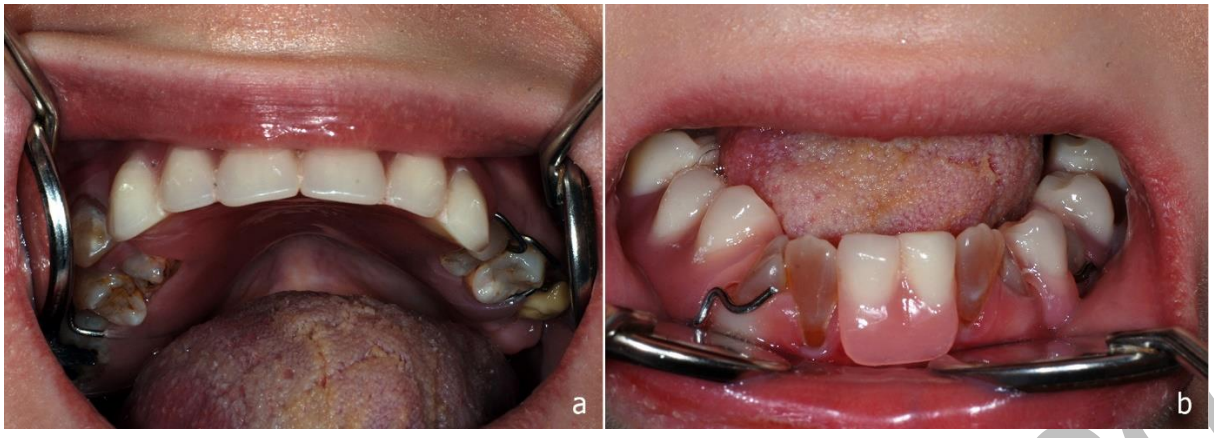

Figure 4. Intraoral photographs with adjusted partial dentures of the: a – maxillary, b – mandibular arch

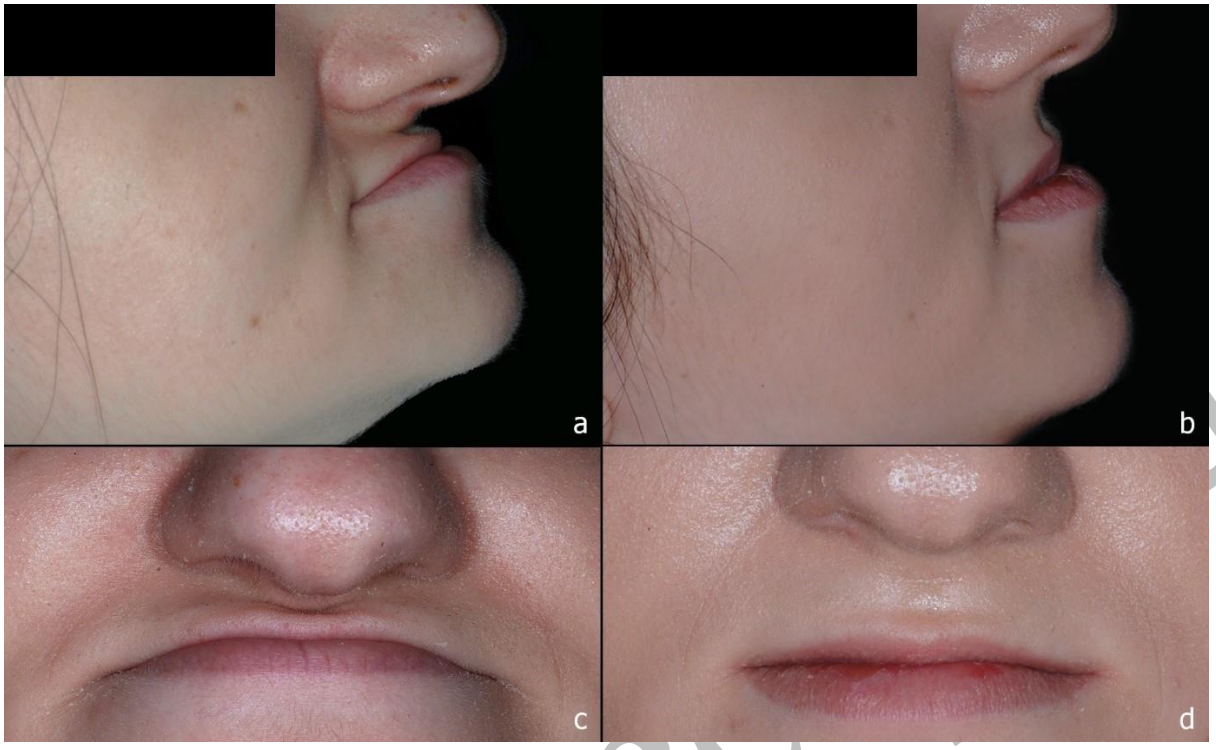

Figure 5. a – Initial extraoral profile view, mandibular prognathism with maxillary hypoplasia; b – facial appearance after prosthodontic rehabilitation and restored vertical occlusal dimension; c – labial philtrum and upper lip before the prosthodontic treatment; d – after the treatment

