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Case Report

Dental Manifestations and treatment of Vitamin D Dependent Type 2 Rickets - A Case Report

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Abstract

Vitamin D–dependent rickets type II (VDDR-II) is a rare autosomal recessive disorder characterized by end-organ resistance to 1,25-dihydroxyvitamin D_3, resulting in defective calcium and phosphate metabolism. Besides skeletal abnormalities, the condition presents with distinctive dental manifestations that are often underrecognized. This case report describes a 17-year-old female with VDDR-II who exhibited delayed tooth eruption, enamel hypoplasia, widened pulp chambers, multiple carious lesions, periapical pathologies, and supernumerary teeth. Clinical and radiographic findings were consistent with rickets-associated dental anomalies. A multidisciplinary treatment plan involving pediatric dentistry and endodontics was implemented, focusing on preventive care, restorative procedures, and necessary endodontic therapy, with special consideration of her systemic status. Oral hygiene reinforcement and periodic follow-ups were emphasized. This coordinated approach resulted in improved oral health and enhanced quality of life. Dental practitioners play a vital role in the early recognition and comprehensive management of systemic disorders such as VDDR-II. Awareness of its oral manifestations facilitates timely diagnosis, reduces complications, and highlights the importance of interdisciplinary collaboration for holistic patient care.

Keywords: Alopecia, Rickets,Vitamin D, Vitamin D Dependent Rickets

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Introduction

Vitamin D is a fat-soluble vitamin essential for calcium and phosphate metabolism, promoting bone mineralization and growth. It is synthesized in the skin upon sunlight exposure and obtained from dietary sources. Deficiency impairs bone development, leading to rickets in children and osteomalacia in adults. [1] Rickets is a pediatric skeletal disorder characterized by defective mineralization of epiphyseal growth plates, resulting in weak bones, pain, growth delays, and deformities. It predominantly affects children aged 6 months to 2 years and is more prevalent in African, Middle Eastern, and Asian regions (10–70%). While national prevalence in India remains undetermined, a 2020 clinical study reported rickets susceptibility in approximately 46% of children aged 0–10 years.[1]

Rickets is broadly classified based on etiology into[1]:

1. Calcipenic Rickets
2. Phosphopenic Rickets
3. Rickets due to inhibited mineralization

Genetic forms of rickets are further divided into[1] :

1. Vitamin D-Dependent Rickets - Type I (VDDR - I) and Type II (VDDR - II)
2. Congenital Hypophosphatemic Rickets - FGF - 23 - dependent and independent hypophosphatemic rickets

General skeletal features include craniotabes, frontal bossing, wide fontanels, bowed legs, and chest deformities like ricketic rosary and pigeon chest. Oral signs include delayed eruption, enamel hypoplasia, increased caries risk, and occasionally supernumerary teeth. A.B. Bindu et al. (2020) highlighted the need for dental evaluation, noting the potential presence of supernumerary teeth in rickets.[1,2]

Neurological complicationsin rickets include tetany, seizures, paresthesia, muscle spasms, cardiomyopathy, hydrocephalus, intracranial hypertension, and craniosynostosis. Early recognition of oral and skeletal signs is crucial, especially in genetic forms like Vitamin D-Dependent Rickets Type II.[1,2]

This case report presents the dental features of a patient with Vitamin D-Dependent Rickets Type II (VDDR-II) managed in our department. It highlights the oral manifestations of this rare metabolic disorder and emphasizes the need for a multidisciplinary approach in diagnosis and treatment planning.

Case Report

A 17-year-old female reported to the Department of Pediatric and Preventive Dentistry with mild, progressively increasing pain in the upper right posterior region for 5–6 months, accompanied by nocturnal, radiating pain aggravated by chewing.She is a medically diagnosed case of Vitamin D-Dependent Rickets Type II (VDDR-II), a rare genetic disorder affecting vitamin D metabolism, associated with alopecia and delayed developmental milestones such as speech and walking. She is under regular medical care for vitamin D dependent type 2 and receives cholecalciferol (60,000 IU), calcitriol (0.25 mcg) four times daily with meals, and a calcium-magnesium-zinc-vitamin D3 supplement in divided doses.Careful monitoring and dose adjustment based on the biomedical profile are essential in cases of VDDR type 2.

A positive family history of consanguineous marriage may be linked to the genetic etiology of her condition.She is the second of two siblings, both female, and the elder sister has not exhibited any systemic diseases or medical conditions. Previously, root canal treatment and stainless steel crown placement were completed irt 46, along with a GIC restoration irt 36.

General physical examination revealed a height of 112.5 cm and weight of 50 kg, both below average for age. Clinical signs included alopecia, frontal bossing (Figure 1 and 2), bowed lower limbs, slurred speech, and waddling gait—indicative of rickets and muscle weakness. Biochemical tests showed elevated serum alkaline phosphatase and low total vitamin D (D2 + D3), confirming a diagnosis of VDDR-II.

Radiographic examination of the wrists (AP and lateral views) showed mild deformity of the radius and ulna (Figure 3), while ankle imaging revealed anteroposterior and lateral bowing of the tibia and fibula (Figure 4). Intraoral examination identified deep dentinal caries in tooth 17 and a faulty restoration in tooth 36 (Figure 5), both tender on percussion, suggestive of symptomatic irreversible pulpitis. A supernumerary tooth was noted in the region of 15–16. An orthopantomogram was utilized to validate the clinical impressions and to assist in the development of a structural treatment protocol (Figure 6).

The treatment plan included endodontic therapy for teeth 17 and 36, followed by stainless steel crowns for long-term protection. Oral prophylaxis and topical fluoride application were performed, with fluoride recommended every 6 months.

Discussion

The term “rickets” likely originates from the German word “wricken,” meaning “twisted.” Cases surged during the industrial revolution due to overcrowding, indoor living, smog, and poor calcium and vitamin D intake,particularly

in urban, low socioeconomic populations.[1-6]

Ranjan et al. (2023), Sunuwar et al. (2021), Soni SS et al. (2008), and Mishra S et al. (1996) reported that the clinical features of VDDR Type II commonly include growth retardation, short stature, rachitic changes such as bowing of the legs, widening of metaphyseal ends, and fractures. Alopecia is also frequently associated with this condition.[4] Amal et al. (2024) noted that previous studies have established a link between vitamin D deficiency (VDD) and various orofacial factors. They reported an increased incidence of VDD in children and adolescents, particularly during winter months, due to reduced sunlight exposure.[7] Akif Demirel et al. (2017) suggested that management of such cases may include prophylacticendodontic treatments, conservative therapy, and tooth extractions, followed by rehabilitative treatment when necessary.[8] An-Qi Liu et al. (2020) stated that since calcium and phosphate ions are essential for tooth mineralization, patients with rickets often present with oral complications. However, the oral manifestations can vary depending on the type of rickets.[9]

The patient presented with symptomatic irreversible pulpitis in the maxillary right second molar and mandibular left first molar. Based on clinical and radiographic evaluation, root canal therapy was done.This case of Vitamin D-Dependent Rickets Type II (VDDR2) with alopecia represents a rare genetic disorder marked by end-organ resistance to 1,25-dihydroxyvitamin D.

Endodontic challenges included managing enlarged pulp chambers and potentially altered root canal morphology, features commonly associated with VDDR2.[8,9] Given the compromised enamel and dentin structure, a stainless steel crown was placed to protect the tooth from further fracture and restore function.Stainless steel crowns are durable and cost-effective, making them a preferred choice for young patients with teeth at high risk of structural failure.[8,9]

Considerations for Future Dental Care - Regular fluoride application (every 6 months), oral prophylaxis (every 6–12 months), and pit and fissure sealants were advised. Ongoing follow-up is essential to monitor RCT success, SSC integrity, and detect complications like root resorption or periodontal issues.[8,9]

Conclusion

This case highlights the need for interdisciplinary care in managing systemic conditions with dental manifestations. Collaboration with pediatricians or endocrinologists is vital, along with Vitamin D supplementation to address VDDR2. Conservative dental management ensures optimal oral function and overall health outcomes.[10]

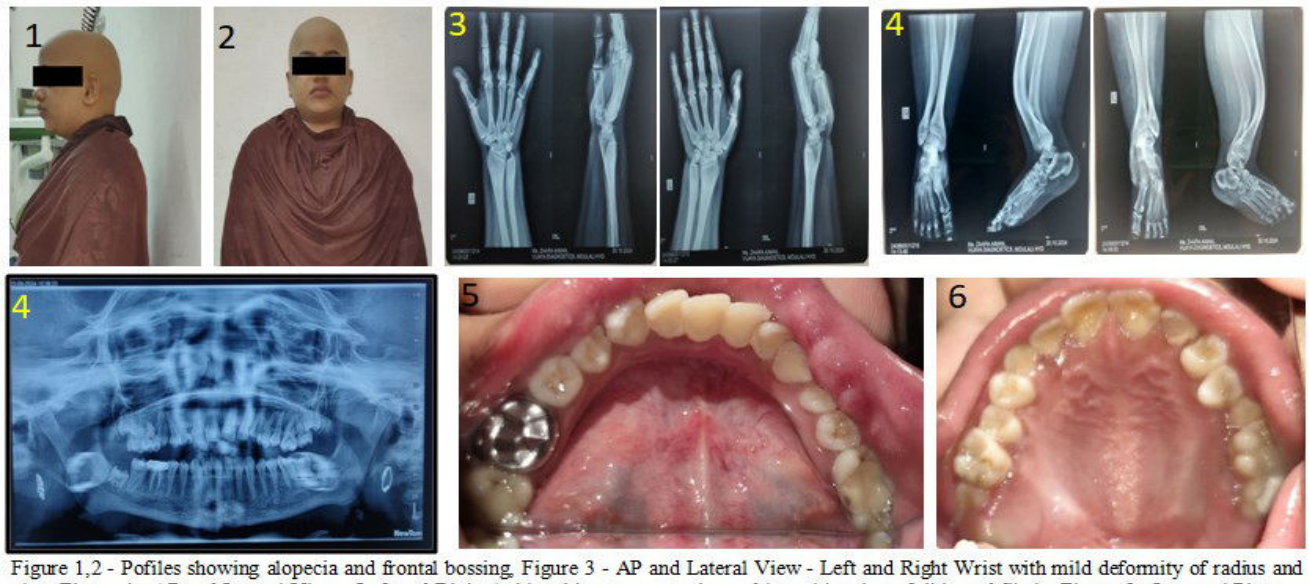

Figure 1,2 - Pofiles showing alopecia and frontal bossing, Figure 3 - AP and Lateral View - Left and Right Wrist with mild deformity of radius and ulna, Figure 4 - AP and Lateral View - Left and Right Ankle with anteroposterior and lateral bowing of tibia and fibula, Figure 5 - Intraoral Pictures showing Deep Dental Caries irt 17, Supernumerary tooth irt 15,16, GIC restoration irt 36 Stainless Steel Crown irt 46, Figure 6 - OPG showing Deep Dentinal Caries irt 17, Supernumerary teeth irt 15,16 Secondary Caries irt 36, RCT treated 46 with stainless steel crown

Clinical Significance

This case report highlights the importance of recognizing the oral and systemic manifestations of rare genetic disorders such as Vitamin D-Dependent Rickets Type 2 (VDDR-II). Early dental findings—such as delayed tooth eruption, enamel hypoplasia, and enlarged pulp chambers—may serve as critical diagnostic indicators, particularly in pediatric patients. Prompt identification by dental professionals can facilitate early medical intervention, reduce long-term skeletal and dental complications, and emphasize the need for interdisciplinary management in improving patient outcomes.

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Conceptualization: MC, Formal Analysis: MC, Investigation:MC, Methodology:MC, Project Administration: MG, Writing – Original Draft:MC,Writing – Review & Editing: KPV

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