DOI: 10.4274/jbuch.galenos.2025.88234
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Case Report

Dental Anomalies in a Pediatric Patient with 16p13.11 Recurrent Microdeletion Syndrome: A Case Report

16p13.11 Rekürrent Mikrodelesyon Sendromlu Çocuk Hastada Dental Anomaliler: Olgu Sunumu

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ABSTRACT

Recurrent 16p13.11 microdeletion syndrome is a rare genetic condition with variable clinical manifestations. This report aims to highlight the craniofacial and dental features of a pediatric patient with this syndrome, emphasizing the importance of early dental evaluation in children with genetic disorders. A 7-year-old girl diagnosed with recurrent 16p13.11 microdeletion syndrome, who had no previous dental examination, was evaluated. Clinical and radiographic findings revealed microcephaly, retrognathic maxilla, Class III appearance, oligodontia, delayed tooth eruption, and malformations in crown and root morphology. Dental caries in molars were restored with glass ionomer, compomer, and composite resin. Due to mental retardation and poor cooperation of the patient, removable prosthetic rehabilitation was not planned. Oral hygiene education was provided, and follow-up visits were scheduled.

This case underlines the significance of early dental assessment in patients with genetic syndromes and emphasizes the need for multidisciplinary management and long-term follow-up.

Keywords: Pediatric dentistry, syndrome, chromosome deletion, oral manifestations

ÖZ

Rekürrent 16p13.11 mikrodelesyon sendromu, değişken klinik özellikler gösterebilen nadir bir genetik durumdur. Bu olgu sunumunda, söz konusu sendroma sahip pediatrik bir hastanın kraniyofasiyal ve dental bulgularının vurgulanması ve genetik bozukluğu olan çocuklarda erken dental değerlendirmenin öneminin ortaya konması amaçlanmıştır. Daha önce hiç dental muayenesi yapılmamış, 16p13.11 rekürrent mikrodelesyon sendromu tanısı bulunan 7 yaşındaki bir kız hasta değerlendirilmiştir. Klinik ve radyografik incelemelerde mikrosefali, retrognatik maksilla, Sınıf III görünüm, oligodonti, gecikmiş diş sürmesi ve kuron-kök morfolojisinde bozukluklar gözlenmiştir. Molar dişlerdeki çürükler cam iyonomer, kompomer ve kompozit rezin ile restore edilmiştir. Zihinsel yetersizlik ve eksik kooperasyon nedeniyle hareketli protetik rehabilitasyon planlanmamıştır. Hastaya oral hijyen eğitimi verilmiş ve takip randevuları planlanmıştır. Bu olgu, genetik sendromu olan hastalarda erken dental değerlendirmenin önemini ortaya koymakta ve multidisipliner yaklaşım ile uzun dönemli takibin gerekliliğini vurgulamaktadır.

Anahtar kelimeler: Çocuk diş hekimliği, sendrom, kromozom delesyonu, oral bulgular

Received: 23.09.2025
Accepted: 18.11.2025
Epub: 03.02.2026

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Cite as: Aksoy H, Ertuğrul F, Özdemir TR. Dental anomalies in a pediatric patient with 16p13.11 recurrent microdeletion syndrome: a case report. J Dr Behcet Uz Child Hosp. [Epub Ahead of Print]

INTRODUCTION

Genetic syndromes are complex disease groups that are frequently encountered in childhood, and often involve multiple organ systems[1]. They may develop due to chromosomal anomalies, monogenic disorders, or submicroscopic genomic alterations such as microdeletions and microduplications[1]. Although their clinical manifestations vary significantly among individuals, neurodevelopmental delay, musculoskeletal abnormalities, dysmorphic facial features, congenital heart defects, and orofacial malformations have been widely reported in pediatric patients[1,2]. Orofacial findings, in particular, may directly, and adversely affect feeding, speech, and psychosocial development, and may also provide important diagnostic clues[2]. Some genetic syndromes also have significant adverse effects on oral and dental health in childhood[2]. For example, individuals with Down syndrome are prone to develop macroglossia, a high-arched palate, delayed tooth eruption, and periodontal disease, while patients with ectodermal dysplasia commonly present with marked hypodontia, conical-shaped teeth, and enamel hypoplasia[3]. Treacher Collins syndrome may affect craniofacial development,

causing cleft lip and palate, mandibular hypoplasia, and associated respiratory complications[3]. In Williams syndrome, wide diastemas, hypoplastic enamel structures, and malocclusions have been reported in addition to the characteristic "elfin" facies syndrome[4]. In most of these syndromes, dental abnormalities appear at early ages and often require long-term planning and multidisciplinary intervention[2].

The recurrent 16p13.11 microdeletion syndrome is a rare genetic syndrome with variable phenotypic features[5]. It is characterized by a 0.8-3.3 Mb deletion located on the short arm of chromosome 16 and involves genes such as *NDE1*, *NTAN1*, *MYH11*, *ABCC6*, *MPV17L*[5]. The *NDE1* gene plays a critical role in the development of cerebral cortex, and deletions involving this gene have been strongly associated with neurodevelopmental disorders, including microcephaly[5]. The *NTAN1* gene is thought to play a role in regulating synaptic activity and the formation of social behaviors[6].

The clinical phenotype of the syndrome may include a wide spectrum of symptoms such as global developmental delay, epilepsy, microcephaly, hypotonia, autism spectrum disorder, attention-deficit hyperactivity disorder, low birth weight, and behavioral disturbances[7,8]. Dysmorphic craniofacial manifestations such as low-set ears, a thin upper lip, a short nose, a wide mouth, micrognathia, and downward-sloping palpebral fissures have also been described[9]. These craniofacial manifestations indicate that orofacial structures are functionally affected, as several genes located within the 16p13.11 region play critical roles in the development of ectodermal-derived tissues[8,9]. Although uncommon, a broad spectrum of dental anomalies has been documented in patients with 16p13.11 microdeletion, including congenital tooth deficiencies (hypodontia and oligodontia), structural enamel defects, malocclusion, and delayed tooth eruption[8,10].

In this case report, we present a 7-year-old girl diagnosed with recurrent 16p13.11 microdeletion syndrome who had no previous dental examination. The aim is to describe the coexistence of oligodontia and multiple developmental dental anomalies, and to emphasize the importance of early multidisciplinary evaluation of patients with similar manifestations.

CASE REPORT

This observational clinical case study. Describes a pediatric patient diagnosed with recurrent 16p13.11 microdeletion syndrome. Because this is a single-patient report, no inclusion or exclusion criteria were applied. Clinical and radiographic examinations, including panoramic imaging, were performed, and medical history, dental findings, treatment planning, and follow-up records were reviewed to ensure diagnostic reliability.

A 7-year-old girl diagnosed with 16p13.11 recurrent microdeletion syndrome was admitted to the Ege University. Faculty of Dentistry, Department of Pedodontics, on February 20, 2025, with the complaint of multiple missing teeth and spontaneous exfoliation of existing teeth.

Review of the patient's medical history revealed that microarray analysis conducted at the Genetic Diseases Evaluation Center on 22 October, 2021 had detected a 1.25 Mb deletion within the 16p13.11 chromosomal region. This deletion involved seven *OMIM* genes including *NTAN1*, *RRN3*, *MARF1*, *MYH11*, *FOPNL*, *ABCC1*, and *ABCC6*. These manifestations were reported as a pathogenic variant consistent with recurrent 16p13.11 microdeletion syndrome. The deletion in the same chromosome region was also detected in the patient's father without any clinical or phenotypic abnormality, and genetic examination of the mother was unremarkable.

Family medical history did not reveal any problems related to the cardiovascular, neurologic, or urinary systems. Extraoral examination revealed developmental delay and a Class III facial appearance. Based on clinical findings and previous medical evaluations, the patient had received the diagnosis of intellectual disability.

Panoramic radiograph obtained on February 20, 2025, showed bilateral congenital absence of the upper and lower permanent lateral incisors, both of lower central permanent incisors, and upper right permanent second premolar (Figure 1). Excluding the third molars, seven permanent teeth were missing, leading to a diagnosis of oligodontia. Intraoral examination confirmed the absence of these teeth in both dental arches.

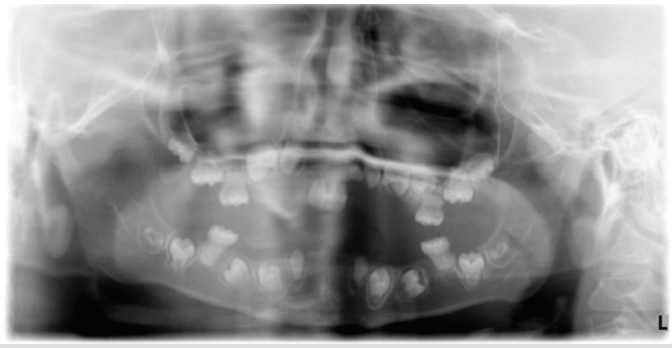

Figure 1. Panoramic radiograph at first admission showing oligodontia and crown-root malformations

According to the parent's report, the anterior teeth in both jaws had previously exfoliated once, and based on this information, the upper right and left central incisors present at admission were interpreted as permanent teeth. Radiographic evaluation also revealed crown and root malformations in several developing permanent tooth germs (Figure 1). In addition, the crown dimensions of these teeth were smaller than normal, consistent with the diagnosis of microdontia (Figure 1). Significant variation was noted in the eruption paths of the tooth germs, and some showed deviation from the anatomical eruption line, with oblique angulation and rotational irregularities (Figure 1).

Intraoral and radiographic examinations demonstrated dentin caries in the upper right and left deciduous second molars, as well as fissure caries in the lower right and left first permanent molars (Figure 1). Increased dental mobility was noted in the maxillary central incisors which was evidenced radiographically by root resorption (Figure 1). Due to the patient's intellectual disability and limited cooperation, intraoral photographic documentation could not be completed during the first visit.

The carious lesion on the upper right deciduous second molar was cleaned with hand instruments, and a glass ionomer restoration was placed to improve patient comfort and relieve pain. The patient was recalled six months later for follow-up. During this visit, cooperation had improved; therefore, detailed extraoral and intraoral examinations could be performed, and intraoral photographs were obtained. Frontal and lateral photographs revealed a prominent Class III facial profile due to retrognathic maxilla and protrusive mandible (Figures 2a, and 2b). Mild microcephaly, a flat nasal bridge, downward-sloping palpebral fissures, a thin upper lip, and low-set ears were also observed (Figure 2a).

In the second intraoral examination, the permanent upper central incisors that had been present at the first visit were missing (Figure 3a). Considering the previously noted advanced mobility and radiographic root resorption, it was concluded that these teeth exfoliated spontaneously. All maxillary anterior incisors were absent. In the maxillary arch, deciduous second molars and permanent first molars were present bilaterally in the posterior region (Figure 3a). Moderate mobility and dentin caries were detected in the upper left deciduous second molar, and fissure caries were found on the permanent molars (Figure 3a).

In the mandibular arch, right and left permanent first molars were present, and fissure caries lesions were observed on their occlusal surfaces (Figure 3b). The right deciduous mandibular canine showed mobility with significant accumulation of dental calculus (tartar) and gingival inflammation (Figure 3b).

Due to the numerous missing teeth and delayed eruption, inadequate development of the dental arches was observed. A "knife-edge" alveolar crest was present in both the maxilla and mandible, characterized by narrow, high, and sharply contoured alveolar ridges (Figures 3a, and 3b).

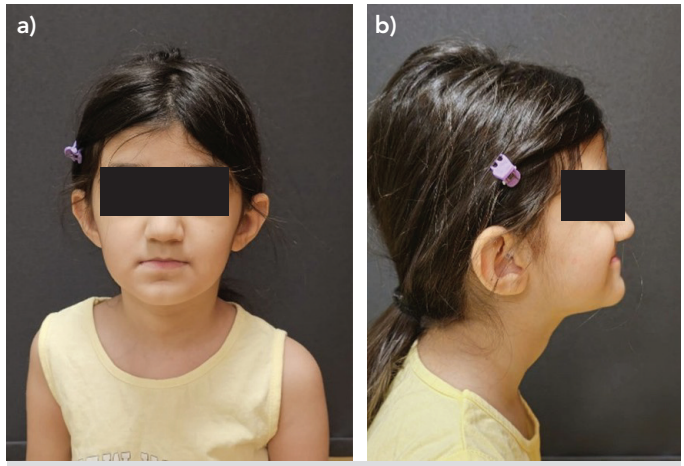

Figure 2. Extraoral views: (a) frontal view with microcephaly and dysmorphic features. (b) lateral view with retrognathic maxilla and Class III profile

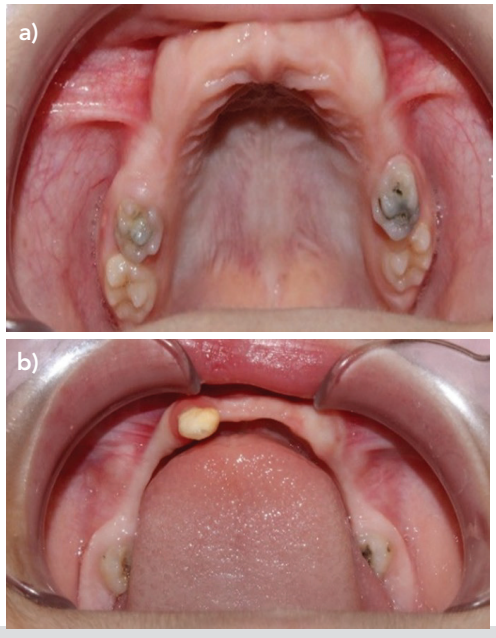

Figure 3. Intraoral occlusal views: (a) missing maxillary incisors and knife-edge alveolar crest. (b) mandible with calculus accumulation and gingival inflammation

The panoramic radiograph obtained during the second visit confirmed these findings. Radiographic examination revealed advanced resorption of the roots of the right deciduous mandibular canine and the left deciduous maxillary second molar, suggesting that spontaneous exfoliation could occur (Figure 4). Similar to the first radiograph, many permanent tooth germs were congenitally absent, and the existing germs were positioned at different angulations with developmental crown and root abnormalities (Figure 4). The crown and root structures demonstrated marked hypoplasia, and the roots were short and conical (Figure 4).

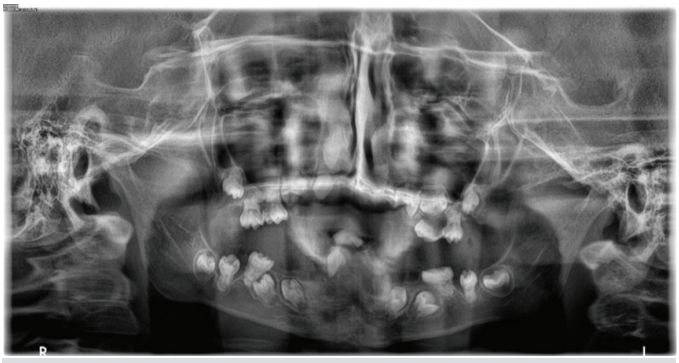

Figure 4. Follow-up panoramic radiograph showing abnormal eruption pathways and root hypoplasia

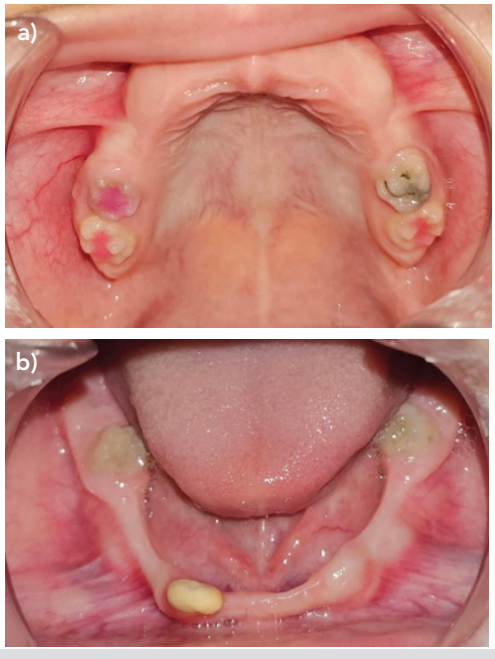

Figure 5. Post-treatment occlusal views: (a) maxilla after compomer restorations. (b) mandible after composite resin restorations

During subsequent treatment sessions, caries in the upper right and left permanent first molars and the upper right deciduous second molar were removed, and the teeth were restored with compomer (Figure 5a). Carious lesions on the permanent mandibular first molars were removed and restored with composite resin (Figure 5b). The upper left deciduous second molar and lower deciduous canine, both demonstrating moderate mobility, were retained under follow-up because the parents did not consent to extraction of these teeth (Figures 5a, and 5b).

Due to the patient's intellectual disability, limited ability to cooperate, and the presence of the narrow, high, and knife-edge shaped alveolar crests, removable prosthetic rehabilitation was not recommended under current conditions. Oral hygiene instructions were provided, and the patient was referred for regular follow-up visits.

DISCUSSION

The recurrent 16p13.11 microdeletion syndrome is a chromosomal disorder that may cause clinical diagnostic difficulties due to its rarity and the considerable variability in genotype-phenotype correlation[7,8]. Similar to the present case, phenotypically unaffected individuals carrying the same deletion have been reported, suggesting that variable expressivity and reduced penetrance may be influenced by environmental or epigenetic modifiers[7].

In the current patient, neurodevelopmental and craniofacial characteristics, including intellectual disability, microcephaly, retrognathic maxilla, and a prominent Class III skeletal profile were overlapped with the features previously described in the literature[8,9]. Dysmorphic facial traits such as a flat nasal bridge, downward-sloping palpebral fissures, a thin upper lip, and low-set ears were also consistent with this syndrome[9]. These clinical findings may assist clinicians in recognizing this syndrome and reinforces the indication for genetic testing in similar cases.

Only a limited number of studies have documented the orofacial manifestations of recurrent 16p13.11 microdeletion syndrome. Dental anomalies such as hypodontia or oligodontia, enamel defects, delayed eruption, and microdontia have been rarely reported in affected individuals[11,12]. In our patient, seven permanent teeth, excluding third molars, were congenitally absent, leading to a diagnosis of oligodontia. Therefore, this case contributes to the literature as a rare example of dental developmental anomalies associated with this chromosomal abnormality.

Radiographic examination clearly demonstrated malpositioned tooth germs, deviations in timing and direction of eruption, rotational anomalies, and developmental defects in crown and root morphology. These abnormalities can adversely affect quality of life not only functionally, but also esthetically and psychosocially. Furthermore, insufficient alveolar bone development due to multiple missing teeth resulted in narrow, high, pointed alveolar crests, which complicated dental prosthetic rehabilitation procedure.

Because of the patient's intellectual disability and behavioral problems, cooperation during dental treatment was limited. As a result, minimally invasive procedures were preferred, and removable prosthetic rehabilitation was postponed. Similar to our findings, previous studies have emphasized that lack of cooperation represents a significant challenge in the dental management of children with neurodevelopmental impairment[13].

In conclusion, this case provides a comprehensive example of the dental manifestations that may accompany 16p13.11 microdeletion syndrome. The coexistence of rarely reported findings, including oligodontia and developmental anomalies in dental morphology, supports the phenotypic diversity of this chromosomal disorder. These results underscore the need for a multidisciplinary approach in pediatric patients presenting with similar characteristics, as early diagnosis and intervention may contribute to improved functional, esthetic, and psychosocial outcomes.

Parents' Perspective

As parents, witnessing the impact of our child's dental condition on her quality of life was a difficult experience. After the completion of the initial treatments, her functional comfort improved, which provided reassurance and optimism for the future. We appreciate the clinical team for their care and remain hopeful about the upcoming prosthetic treatment.

Ethics

Informed Consent: Written informed consent was obtained from the patient's parents for publication of this case report and the accompanying clinical images.

Footnotes

Authorship Contributions

Surgical and Medical Practices: H.A., Concept: H.A., F.E., Design: H.A., F.E., T.R.Ö., Data Collection or Processing: H.A., F.E., T.R.Ö, Analysis or Interpretation: H.A., F.E., T.R.Ö, Literature Search: H.A., Writing: H.A., F.E.

Conflict of Interest: There is no conflict of interest between the authors.

Financial Disclosure: The authors declared that this study received no financial support.

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