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Evaluatıon of Oral Fındıngs and Treatment Approach of a Child Patient wıth Wıllıams Syndrome: Case Report

SUMMARY

Pediatric dentists may encounter patients with rare syndromes. The intraoral and extraoral findings in these patients can vary widely, ranging from mild to complex, depending on the extent of syndrome involvement. It is essential to be knowledgeable about this special needs patient group in order to raise awareness regarding their oral health care requirements.Williams syndrome results from the deletion of a set of genes on chromosome 7. The condition is characterized by distinct facial features, dental anomalies, cardiac issues, hypercalcemia, orthopedic problems, and learning disabilities, which are commonly associated with the syndrome. The aim of this case report is to present the intraoral findings and dental anomalies observed in a pediatric patient with Williams syndrome, as well as to share the treatment approach applied to the patient.

Keywords: Chromosome; Syndrome; Dentistry, Malocclusion.

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CASE REPORT (CR)
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Introduction

Williams syndrome (WS), also known as Williams-Beuren syndrome, is an autosomal dominant disorder. Although the exact incidence is unknown, it is estimated to occur in approximately 1 in 20,000 live births. The syndrome has been identified in all ethnic groups worldwide and affects both sexes equally [1]. Individuals diagnosed with WS have a de novo deletion of 1.5 to 1.8 Mb at 7q11.23, and although rare, an affected parent of a person with WS may also carry this deletion [2]. This syndrome is a multisystem disorder that affects both mental and physical development. The deleted region of chromosome 7q11.23 observed in WS includes the ELN gene, which encodes the elastin protein, a critical component of elastic fibers found in the connective tissue of many organs[3–6].

WS is characterized by distinctive facial features (elfin-like face), cardiovascular anomalies, primarily supravalvular aortic stenosis (SVAS), growth delay, early puberty, endocrine disorders such as hypothyroidism, intellectual disability with a typical neurobehavioral profile, and often infantile hypercalcemia[5,7].

Additionally, facial features include a broad forehead, bitemporal narrowing, periorbital fullness, a stellate/lacy iris pattern, strabismus, a short nose with a broad nasal tip, malar flattening, a long philtrum, thick vermilion line of the upper and lower lips, and a wide mouth[2].

The identified intraoral findings of the syndrome include generalized diastemas (70%), a high-arched palate, hypodontia (50%), microdontia, and enamel hypoplasia[8,9].

Additionally, these patients may present with malocclusion conditions such as small roots, dens invaginatus, anterior crossbite, anterior deep bite, or open bite[5]. Dental malocclusion is present in 85% of individuals with WS and responds to orthodontic treatment. Poor fine motor skills pose challenges in maintaining oral hygiene and increase the risk of dental caries[2].

The aim of this case presentation is to describe the facial, oral, and dental features observed in a pediatric patient with Williams Syndrome and to detail the therapeutic approaches employed.

Case Report

A 13-year-old female patient presented to our clinic with a complaint of pain in the lower left first molar. It was learned from the medical history that she was diagnosed with Williams Syndrome at the age of 5 and is not currently taking any medication. The patient was noted to be attending a special education center.

During the extraoral examination, significant findings included periorbital fullness, a broad forehead, an elongated philtrum, a thick vermilion border of the upper and lower lips, and a wide mouth (Figure 1).

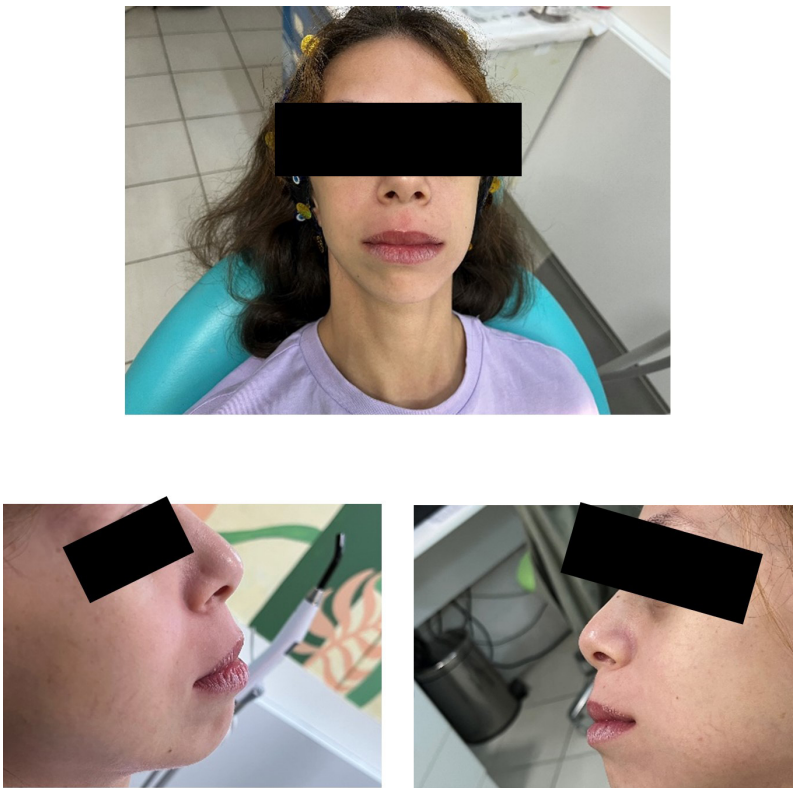

Figure 1: Front and profile view of the patient

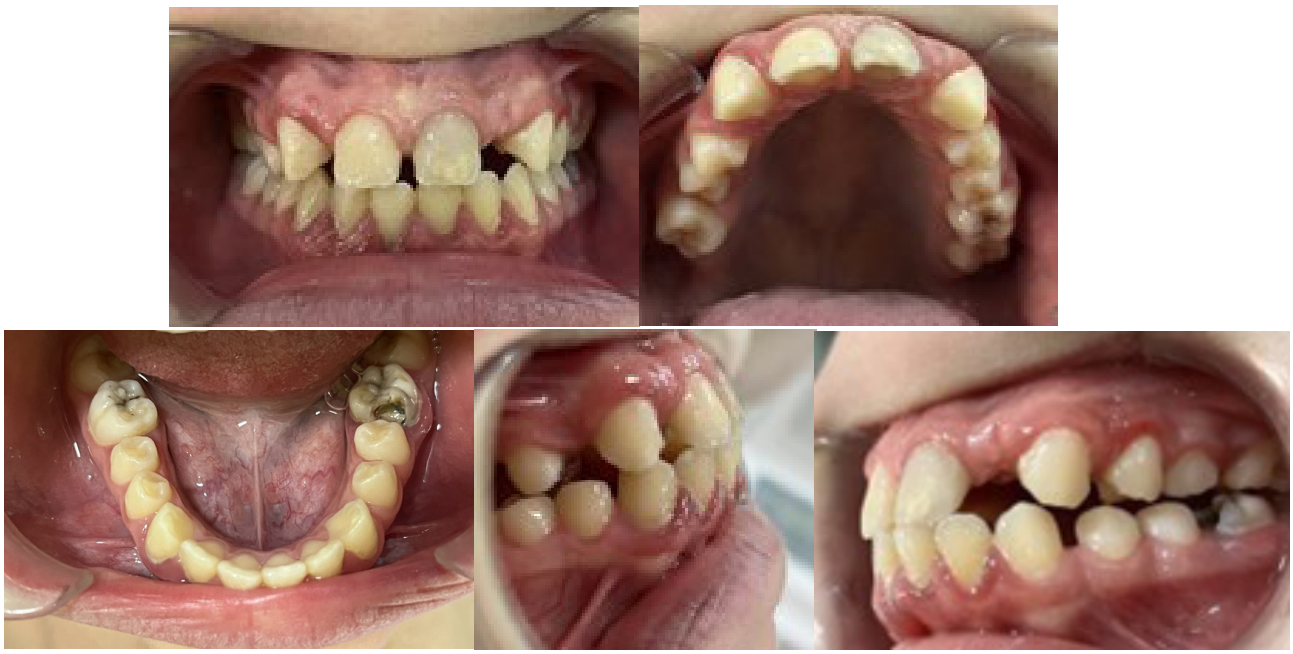

Figure 2: intraoral images of the patient

It was noted that 10 days prior, the patient developed swelling in the lower left first molar region, causing facial asymmetry, for which antibiotics were prescribed.

In the intraoral examination, a deep carious lesion involving the pulp was detected in tooth number 36. Swelling was observed in the buccal mucosa at the gingival margin of the affected tooth. Localized calculus was identified, particularly around the anterior teeth of the lower jaw. It was noted that teeth numbers 16, 12, and 22 were absent in the oral cavity. Tooth number 16 had been extracted when the patient was 8 years old, and teeth numbers 12 and 22 had never been present. Occlusal caries were found in teeth numbers 46 and 26. A hypomineralized area was observed on the facial surface of tooth number 21. When evaluating the relationship of the jaws during occlusion, it was noted that the patient exhibited maxillary protrusion and had a end-to-end occlusion. Widespread diastema was observed in the maxilla (Figure 2).

Radiographic examination revealed the presence of radiolucency in the periapical region of tooth number 36, including the furcation area. Additionally, it was determined that teeth numbers 12, 22, 18, and 28 were congenitally missing (Figure 3).

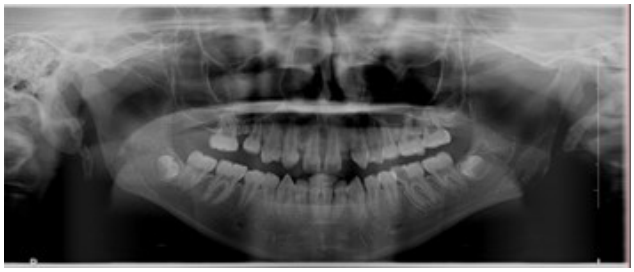

Figure 3: panoramic radiograph of the patient

Treatment Planning

In the treatment planning for the patient with Williams Syndrome, cardiac issues were investigated, and it was determined that there were no existing problems. Nevertheless, a consultation with cardiology was conducted to assess the need for prophylaxis, and it was reported that cardiac prophylaxis was not necessary.

Since the patient exhibited severely limited cooperation and was unable to tolerate prolonged dental treatment in the chair, extraction of tooth number 36 was planned with the approval of the guardian. Following the application of appropriate behavioral guidance techniques, the tooth was extracted in a single session using a non-traumatic approach. No postoperative complications were observed. Dental cleaning was performed, and restorations were completed for teeth numbers 46 and 26. The patient was referred to the orthodontic department due to the presence of malocclusion and missing teeth. It was decided to monitor the hypomineralized area on the facial surface of tooth number 21. In addition, an individualized preventive oral care plan was established, including oral hygiene instructions for both the patient and parent. Emphasis was placed on twice-daily fluoride toothpaste use, mechanical plaque control, and interdental cleaning. The routine dental visits at three-month intervals were advised. Given the patient's high caries risk, topical fluoride applications were planned for follow-up appointments.

Discussion

Williams Syndrome (WS) is characterized by distinctive facial features (elfin face), cardiovascular anomalies such as supravalvular aortic stenosis (SVAS), growth delay, intellectual disability with a typical neurobehavioral profile, and often infantile hypercalcemia[10,11].

The literature indicates that individuals with Williams Syndrome possess characteristic elfin facial features[2]. During the extraoral examination of our patient, prominent features of the syndrome were observed, including a broad forehead, periorbital fullness, an elongated philtrum, and a wide vermilion border.

It has been reported that a significant portion of patients with Williams Syndrome have supravalvular aortic stenosis (SVAS). This condition may necessitate the administration of antibiotic prophylaxis prior to dental treatment[11]. Therefore, our patient was referred to a cardiology specialist, and it was determined that prophylaxis was not necessary.

Intellectual disability is observed in a significant portion of individuals with Williams Syndrome. However, our patient demonstrated limited cooperation during the examination and treatment process. The periapical lesion associated with tooth number 36, which was the primary reason for the clinic visit, necessitates a lengthy and multi-session endodontic treatment. Considering the patient's restricted collaboration, it was decided to extract tooth number 36 with the guardian's approval.

Among the intraoral findings of the syndrome, there are dental anomalies, including tooth deficiencies[12]. In this case, both clinical and radiological examinations revealed that teeth numbers 12, 22, 18, and 28 were congenitally absent. In cases of hypodontia due to Williams Syndrome, multidisciplinary treatment approaches are recommended to eliminate aesthetic and functional losses. In this context, it is possible to control missing tooth gaps with orthodontic treatment or to prepare space for prosthetic restorations. Regarding prosthodontics; while temporary solutions such as tooth-attached placeholders and fiber-supported composite restorations can be offered in growing individuals,

fixed bridges or implant-supported restorations may be preferred after the completion of skeletal development[13,14] .However, as in this case, the syndrome-induced mental limitation and lack of cooperation limit the applicability of these treatments in the short term. Therefore, the recommended orthodontic and prosthodontic approaches should be considered at the planning stage and reevaluated when the patient's level of cooperation improves.

Dental hypoplasia is another intraoral finding associated with the syndrome[9]. Hypoplasia of the enamel was observed on the buccal surface of the patient's tooth number 21. It was decided to monitor the related tooth during follow-up appointments.

Conclusions

It is difficult to state that there is a standard treatment protocol for Williams Syndrome from a dental perspective. The primary approach to treatment is symptom-oriented. Individuals with Williams Syndrome require regular monitoring for their medical and dental issues. The most crucial step in the management of these patients is to educate their caregivers about oral and dental health.

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