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Case report

WILLIAMS-BEUREN SYNDROME- A CASE REPORT

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ABSTRACT

Williams syndrome is a rare neurodevelopmental disorder caused by the spontaneous deletion of genetic material from the region q11.23 of one member of the pair of chromosome 7, so that the person is hemizygous for those genes. Syndrome is a rarely genetic multisystem disorder that occurs equally in all ethnic groups and both sexes. Williams syndrome is a developmental disorder that affects many parts of the body. This condition is characterized by mild to moderate intellectual disability or learning problems, unique personality characteristics, distinctive facial features, and heart and blood vessel (cardiovascular) problems. The diagnosis of this syndrome is based on recognition of the characteristic pattern of dysmorphic facial and physical features. The diagnosis is typically suspected based on symptoms and confirmed by genetic testing. Treatment includes special education programs and various types of therapy. Surgery may be done to correct heart problems. Dietary changes or medications may be required for high blood calcium. Life expectancy is less than that of the general population, mostly due to the increased rates of heart disease. This case report presents the story of 10-year-old girl who suffers from Williams syndrome. Cardiac murmur, mental retardation, severe malocclusion problems, wide mouth, diestama, full lip, periorbital fulness, epicanthal fold, stellate iris pattern, short stature, weaknesses in retional/conceptual language and short term memory observed in this patient. In this study, patient's salivary glands and deciduous anterior tooth were investigated by detailed diagnostic methods (USG, micro-CT). The overall purpose of this case was to diagnose dental manifestations and evaluate physical and clinical characteristics of present case with Williams Syndrome.

Key words: Williams Syndrome, salivary glands, tooth, deciduous, heart diseases.

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INTRODUCTION

Williams (Williams-Beuren) syndrome (WS) is a complex systemic disorder that relevant a hemizygous microdeletion of chromosome 7q11.23, encompassing the elastin gene.[1] This syndrome characterized by supravalvular aortic stenosis, distinctive facial features, growth deficiency, mental retardation and learning disabilities.[2-7] This rare congenital disorder was first described in 1961.[2] Williams *et al.* called attention to a syndrome of supravalvular aortic stenosis, mental retardation, and distinctive facial features. Then in 1962, Beuren *et al.*[8] expanded it to include peripheral pulmonary and dental anomalies.

In the great majority of cases, WS is a sporadic event of unknown cause.[9] There are limited data in the literature on syndromes prevalence in the general population but is estimated to be 1 in 20,000 live births. Both sexes affected equally.[10] The diagnosis is based on recognition of the characteristic pattern of dysmorphic facial features, short stature, developmental delay, connective tissue abnormalities affecting the cardiovascular organs, unique cognitive profile, learning difficulties and sometimes transient infantile hypercalcemia.[10,11] A seriously high proportion of this syndrome was reported to have congenital heart disease.[10] WS phenotype consists of a distinct pattern of physical, behavioral, neurologic and cognitive abnormalities evolving from early childhood.[10,1]

This study aimed to evaluate dental manifestations and physical characteristics of the present case with WS.

CASE REPORT

The procedure performed was in accordance with the ethical standards of the institution and with the 1964 Helsinki declaration and its later amendments. Informed consent was obtained from the patient included in the study and additional written informed consent was obtained from the patient included in the study and additional written informed consent was obtained from the patient for publication of this case report and the accompanying images.

This paper presents a 10-year-old girl who suffers from WS that referred to Ankara University Faculty of Dentistry because of persistent deciduous teeth and routine control. The medical history of the present case showed that the patient had a heart murmur.

In clinical examination; persistent deciduous teeth #51, malpositioned permanent teeth #11 and #21, early eruption, diastema at the front region, severe malocclusion problem, mandibular prognathism, U-shaped arc structure at mandibula (Figure 1) and maxilla (Figure 2), soft tissue hyperplasia and wide mouth structure were detected.

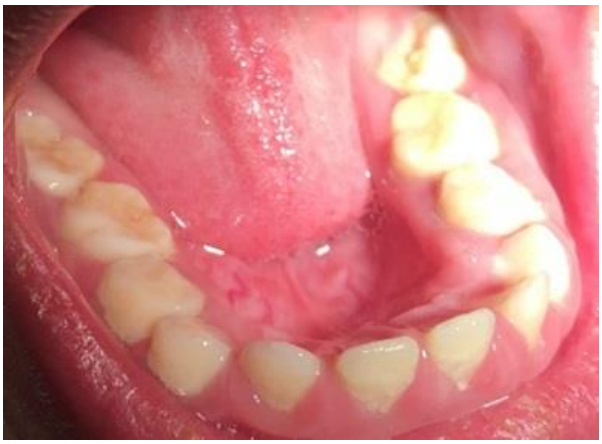

Figure 1. U shaped arc and diastema at the front region intraoral image

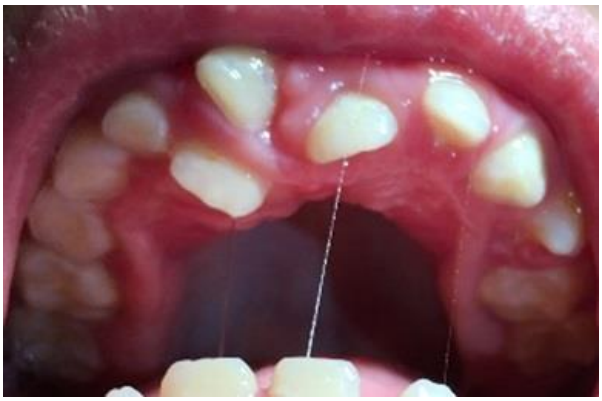

Figure 2. Persistent deciduous teeth and malpositioned permanent tooth intraoral image

Also, plaques on tooth surfaces were detected. But there was no decay, white spot lesion and even fissure coloration detected (dmft 0). There was no intervention at the first appointment, parents just informed about oral hygiene practice. One month later, teeth #51 was extracted. Then, removed tooth and one noncarious extracted tooth #51 from a healthy patient was compared with micro-CT (Figure 3 and Figure 4).

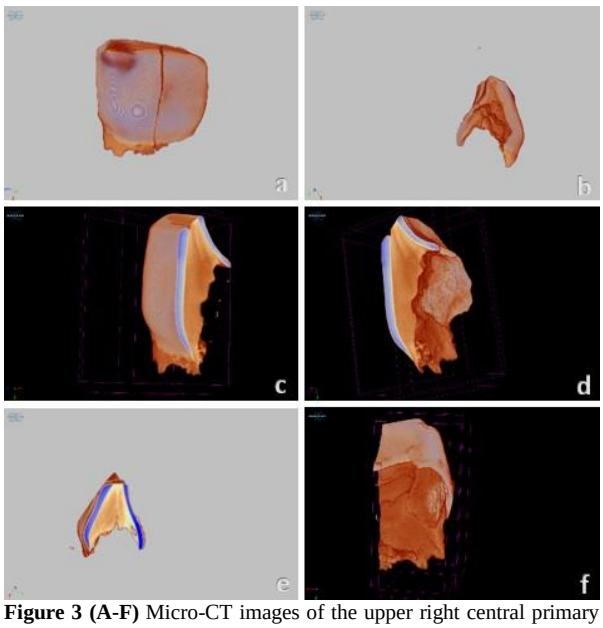

Figure 3 (A-F) Micro-CT images of the upper right central primary incisor of the present case. (A) front section of tooth, (B-E) saggital section of tooth, (F) palatinal section of tooth

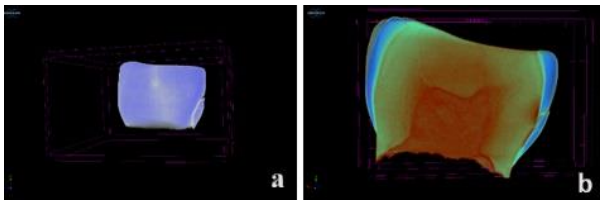

Figure 4 (A, B) Control group's teeth #51 micro-CT image. (A) front section of tooth, (B) vertical section of tooth

According to micro-CT results, the pulp chamber of the patient with WS was detected wider and thinner than the control tooth. Then, the patient was requested to have USG to evaluate the salivary glands., Bilateral parotid and submandibular glands were examined in this examination. Size and contours of bilateral parotid glands were normal. The parenchyma of the glands was homogenous mostly but with heterogeneous foci in some regions of the superficial lobe. Intraparotideal lymph nodes were also observed with a dimension of 1x3 mm in both parotid gland. The dimensions of the bilateral submandibular glands were fairly hyperplasic, but contours were normal. The parenchyma was heterogeneous in both glands. There was no solid or cystic lesion detected in the parenchyma. Especially in the left submandibular gland vascularisation get increased. On both sides, there is a reactive lymph node which is enlarged in the submandibular region.

DISCUSSION

WS's distinctive facial features are; microcephaly, a broad forehead, medial eyebrow flare, periorbital fullness, strabismus, stellate iris pattern, flat nasal bridge, malar flattening, along small philtrum, full cheeks and lips a rather pointed chin and a wide mouth. The face becomes more coarse with age.[12]

Syndrome's distinctive physical features reported in some cases are; mild degrees of short stature, kyphoscoliosis, a long neck, inguinal or umbilical herniae, small nails and hallux valgus.[1] From these distinctive features; short stature, a broad forehead, medial eyebrow flare, periorbital fullness, strabismus, stellate iris pattern, pointed chin observed in this case.

Many studies showed that some of the children had mandibular prognathism, temporomandibular joint abnormalities and dental malocclusion as in this case, but did not consider these findings part of the syndrome.[2,13]

Beuren *et al*.[3] reported that two male children demonstrated agenesis of deciduous and permanent teeth, bud-shaped deciduous maxillary molars, small permanent incisors, broad maxillary and small mandibular dental arches with bilateral posterior scissors-bite. And emphasize that the girls had similar but less pronounced dental malformations. But in this case; tooth structure and size differences, agenesis, small mandibular dental arc with posterior scissors-bite were not observed.

The most frequently observed tooth agenesis pattern seems within the three groups; incisors, canines/premolars, and molars. The general dental agenesis pattern seems to be associated with individual innervation pathways.[14] Despite all this, agenesis has not been observed in this case Some studies have reported some increased frequency of dental abnormalities like hypodontia, microdontia, invagination of maxillary incisors, small and slender roots, increased space between teeth, enamel hypoplasia, high prevalence of dental caries and malocclusion.[10,15,16] These findings were generally reported in medical reviews of limited numbers of WS cases. Except for malocclusion and diastema, no other findings are observed in this case.

45 individuals with WS aged 13 months to 28 years, giving prevalence rates of different dental findings of tooth abnormalities, dental carries and

malocclusion traits.[17] In the other study clinical and radiological examination of 37 individuals with WS 2 to 24 years, it reported that, caries in the deciduous dentition, hypodontia and mineral content in deciduous incisors.

Craniofacial features have reported as dolichocephaly, bitemporal depressions, asymmetry, full cheeks, periorbital fullness, epicanthal folds, stellate or lacy iris patterns, full nasal tip, full lips, long philtrum, wide mouth, and dental malocclusion.[10] Also, periorbital fullness, epicanthal fold, stellate iris pattern, full lip, wide mouth, malocclusion were observed in this case.

In some cases, WS patients have cardiovascular problems which include supravalvular aortic stenosis; failure to thrive in infancy; transient neonatal hypercalcemia.[4,18-20] Hypercalcemia is an infrequent feature when a large series of cases are reviewed.[21] But supravalvular aortic stenosis and arterial narrowing are frequently present.[22] In this case, the only cardiac murmur was observed systematically.

Valve involvement may add the murmur of aortic incompetence to the systolic murmur transmitted to the carotids.[23]

As seen in this case, delayed language and abnormal sensitivities to classes of sounds (hyperacusis) can be observed in the WS.[24,25] They use complex expressive language and they usually have a hoarse voice.[26,27]

WS patients have variable mental retardation. Our patient shows moderate mental retardation. Most patients have mental retardation with an IQ between 50 and 70.[11,28] William syndrome patients usually have a friendly, outgoing personality. Also, our patient was so extrovert and social. According to family history, genetic transition not observed in this case. Already it is stated that WS is rarely familial.[27]

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CONFLICTS OF INTEREST STATEMENT

None

Williams-Beuren Sendromu- Vaka Raporu

ÖZ

Williams sendromu 7q11.23 kromozomunun silinmesinden kaynaklanan, tüm etnik gruplarda ve her iki cinsiyette eşit ve nadir olarak gözlenen nörogelişimsel multisistemik bir hastalıktır. Vücudun birçok bölümünü ilgilendiren gelişimsel bir hastalık olan sendrom, hafif ile orta derecede zihinsel engellilik veya öğrenme problemleri, farklı kişilik özellikleri, kendine has yüz özellikleri ve kardiyovasküler problemler ile karakterize edilir. Sendromun tanısı, karakteristik dismorfik yüz özellikleri ve karakteristik fiziksel özellikler ile mümkündür. Tanılar, genetik testlerle onaylanır. Tedavi, özel eğitim programlarını ve çeşitli terapileri içerir. Kardiyovasküler problemleri düzeltmek amacıyla ameliyat yapılabilir. Bu hastalarda yüksek kan kalsiyum değeri gözlenebildiği için diyet değişiklikleri veya ilaç tedavisi de diğer tedavi seçenekleridir. Yaşam beklentisi, çoğunlukla kalp hastalığı oranlarının artması nedeniyle genel popülasyondan daha azdır. Bu olgu sunumu, Williams sendromu teşhisi konulmuş 10 yaşındaki kızın öyküsünü sunmaktadır. Bu hasta; kardiyak üfürüm, zekâ geriliği, ciddi maloklüzyon problemleri, geniş ağız, diestama, periorbital dolgunluk, epikantal kıvrım, stellat iris paterni, kısa boy, konuşmada zayıflıklar ve kısa süreli hafızaya sahiptir. Bu çalışmada, hastanın tükürük bezleri USG ile ve süt ön dişleri micro-CT ile ayrıntılı olarak incelenmiştir. Bu olgunun genel amacı, sendromun diş hekimliği ile ilgili belirtilerini teşhis etmek ve mevcut vakanın fiziksel ve klinik özelliklerini değerlendirmektir. ***Anahtar Kelimeler:*** *Williams sendromu, tükürük bezleri, diş, süt dişi, kalp hastalıkları.*

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