OLGU SUNUMU CASE REPORT

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Multiple Unerupted and Supernumerary Teeth in a Patient with Williams-Beuren Syndrome: An Unusual Case

Williams-Beuren Sendromlu Bir Hastada Multipl Sürmemiş Sürnumere Dişler: Nadir Bir Olgu Sunumu

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ABSTRACT Williams-Beuren syndrome (WBS) is a genetic disorder, caused by a deletion of chromosome 7. WBS characterized by typical facies and personality, cardiovascular disease, connective tissue growth and endocrine abnormalities. In the oral cavity, malocclusion, micrognathism, hypodontia, enamel hypoplasia, microdontia are among the wide spectrum of abnormalities identified in this disease. In this study, orofacial features of a 14-year-old male patient with WBS were presented. This case, contrary the literature, had multiple supernumerary teeth. The literature was reviewed because of this rare condition.

Keywords: Congenital abnormalities; tooth abnormalities; Williams Beuren syndrome

ÖZET Williams Beuren sendromu (WBS) kromozom 7'de mikrodelesyonların neden olduğu genetik bir bozukluktur. WBS kardiyovasküler hastalık, karakteristik fasiyal ve kişilik özellikleri, bağ dokusu büyümesi ve endokrin anormallikleri ile karakterizedir. İntraoral olarak maloklüzyon, mikrognati, hipodonti, mine hipoplazisi ve mikrodonti tespit edilen geniş anomali yelpazesi arasındadır. Bu çalışmada, WBS'li 14 yaşındaki bir erkek hastanın orofasiyal özellikleri sunulmuştur. Bu olguda, literatürün aksine, multipl sürnumere dişler mevcuttur. Nadir görülen olgu hakkında literatür bilgileri yeniden gözden geçirilmiştir.

Anahtar Kelimeler: Diş anomalileri; doğumsal anomaliler; Williams Beuren sendromu

Williams-Beuren syndrome is characterized by cardiovascular disease, typical facies and personality, mild mental retardation, unique personality, connective tissue growth and endocrine abnormalities.[1] WBS is caused by a 1-2 Mb microdeletion in 7q11.23, a section that includes 28 genes.[2] The ELN and GTF2I genes are located on chromosome 7 in this region.[3,4] Homozygous loss of GTF2I results craniofacial abnormalities, growth retardation and neurologic abnormalities.[5,6] Deletion in the ELN gene is the cause of cardiovascular disease in the syndrome. The etiology is unknown and most cases occur sporadically. Although familial cases with autosomal dominant inheritance have been reported.[7] The prevalence is estimated to be one in every 7,500 live births.[8] The disorder occurs equally in both genders. There is no ethnic or racial discrimination.[9] In the oral cavity, broad mouth, malocclusion, micrognathism, hypodontia, enamel hypoplasia, microdontia, short roots, excessive interdental space, pulp stones and abnormal tooth morphology are

among the wide spectrum of abnormalities identified in this disease.[9-11] Conventionally, diagnosis of WBS is made on the basis of clinical assessment but definite diagnosis is generally established at birth with the fluorescent in situ hybridization (FISH) test. Although facial deformities and dental anomalies are common in patients with WBS, the oral abnormalities are considerably varied and have received limited consideration in the literatüre.[9,12] The purpose of this case report is to identify the distinctive facial and uncommon oral findings of WBS.

CASE REPORT

Parental written informed consent was obtained for case report and disclosure of child's uncovered photographs. A 14 year old male patient presented to dentomaxillofacial radiology clinic, with a complaint of upper right canine tooth position. Patient's medical history revealed underlying Williams-Beuren syndrome. Family history of the syndrome was absent. Therefore, sporadic inheritance is observed. There is a congenital cardiac defect in the patient but not treated, only controlled. Auditory hyperesthesia and mental retardation not present. Detailed history, extra-oral examination, intra-oral examination, and radiographic evaluations were performed. The patient's height was 1.42 cm and the weight was 37 kg. Extra-oral examination revealed malar hypoplasia, thick lower lip, broad mouth, extended philtrum, short nose and bitemporal narrowing, protrusive ear lobes and widely spaced eyes. The characteristic "elfin facies" seen in WBS was noticed (Figure 1). Intraoral examination revealed upper narrow arch, dental midline right deviation and tooth crowding. 11,14,15,16,25,26,27,35,37, 44,45,47 were decay. Plaque and calculus deposits, especially in the mandibular anterior region, were noticed (Figure 2, Figure 3). Panoramic radiograph revealed taurodontism in molar teeth, enlargement of periodontal ligament space of teeth and impacted many supernumerary teeth in all quadrants. As per the panoramic radiograpy, ten supernumerary teeth could be rule out in total; six in maxilla and four in mandible (Figure 4).

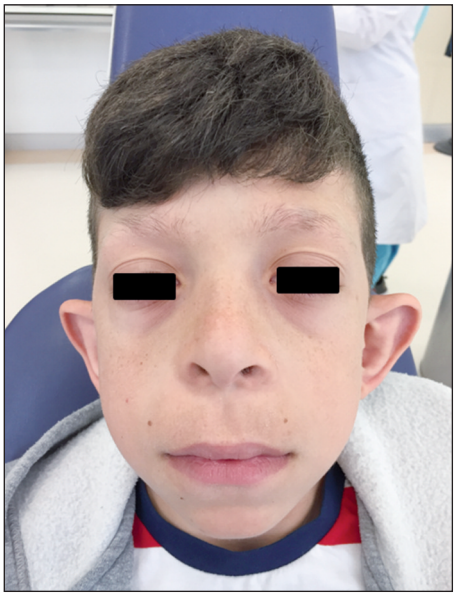

FIGURE 1: Extra-oral image shows thick lower lip, extended philtrum, short nose and broad mouth, bitemporal narrowing, protrusive ear lobes and widely spaced eyes.

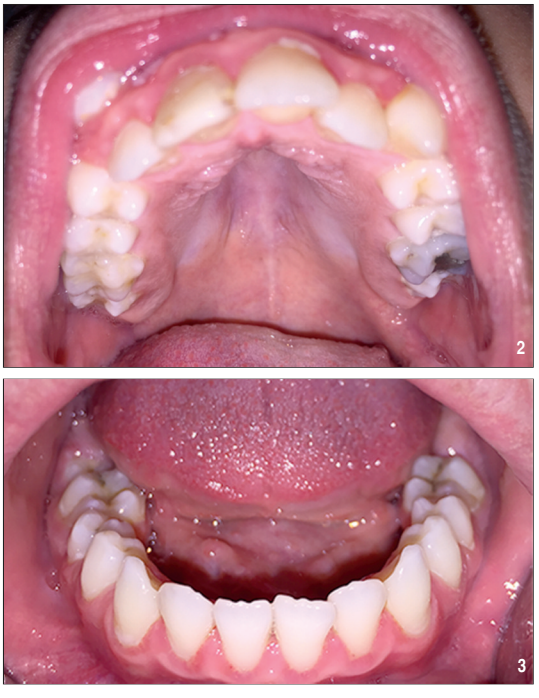

FIGURE 2, 3: Maxillary intraoral image shows the right upper canine tooth in labial position.

DISCUSSION

WBS is caused by a 1-2 Mb microdeletion in 7q11.23, a section that includes 28 genes.[2] Most cases occur sporadically and this patient also has

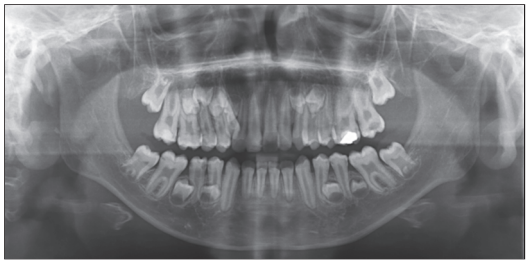

FIGURE 4: Panoramic radiography shows taurodontism in molar teeth, enlargement of periodontal ligament space of teeth and impacted many supernumerary teeth in all quadrants.

sporadic inheritance. WBS should be distinguished from Noonan syndrome, DiGeorge syndrome, Kabuki syndrome, Smith-Magenis syndrome and fetal alcohol syndrome. WBS is characterized by supravalvular aortic stenosis and elastin arteriopathy, distinctive facial findings (e.g., periorbital fullness, broad forehead, malar flattening, extended philtrum, thick vermillion of lips and protrusive ear lobes), auditory hyperesthesia, hoarse voice, mental retardation, talkative personality with lack of stranger anxiety, specific phobias, connective tissue abnormalities and endocrine disorders.[13,14] Computed tomographic angiography (CTA) is an appropriate and noninvasive method for determining the degree of arthropathy associated with deletion in the ELN gene.[15] Magnetic resonance imaging (MRI) is used to demonstrate neuroanatomical and vascular abnormalities specific to WBS.[16,17] Endocrine disorders include hypercalcemia, glucose tolerance, diabetes mellitus and thyroid disfunction.[18] Lateral cranial x-ray showed increased sella turcica size.[19] This may be important in terms of neurological and endocrinological features. The short stature is common but not severe.[20] In this report, the patient has a congenital heart defect, dysmorphic facial features and short stature.[21] In the oral cavity broad mouth, malocclusion, micrognathism, hypodontia, enamel hypoplasia, microdontia, short roots, excessive interdental space, pulp stones and abnormal tooth morphology are among the wide spectrum of abnormalities identified in this disease.[9-11] Cingano et al. detected a higher prevalence of gingival and periodontal infections in the WBS patients.[22] The researchers describe this reason by the poor oral hygiene routines, cooperation deficiency of the young patients and association with the malocclusion.[22] He has broad mouth, malocclusion, a high prevalence of dental caries and enlargement of periodontal ligament space of teeth. Some researchers investigated 45 patients with WBS and found hypodontia in 11.1% of the patients, abnormal tooth morphology was detected in 40.7% of the permanent teeth and 12.5% of the deciduous teeth.[9] Axelsson et al. examined that 52.4% of the patients had agenesis of one or more permanent teeth.[23] Cingano et al. checked four patients with WBS. In all patient the agenesis of two or more teeth were detected, cross bite, deep bite and open bite malocclusion were present too.[22] According to this studies, In WBS cases, hypodontia is common. Contrary to literature, he has supernumerary teeth. Different gene deletions have been reported in patients with WBS. The presence of supernumerary teeth in this case may have been the result of a different genetic deletion than the one reported so far. More genetic studies should be done to determine the cause of this condition. Differently from literature he has taurodontism in molar teeth. Dental infections may increase the risk of developing bacterial endocarditis. Enamel defects are frequent in patients with WBS and tooth loss if initially treatment is not done. When dental methods are required, the management of antibiotics is recommended as treatment for prophylaxis for bacterial endocarditis. Individuals with Williams syndrome are hypersensitive to certain frequencies of sound (auditory hyperesthesia).[24] Thus, any form of dental equipments such as ultrasonic scalers, highspeed handpieces, which generate sound should be first demonstrated, before being used on the patient.

The present case report was found great numbers of orofacial abnormalities in patient with WBS. Previously unreported dental abnormalities have been described in the literature. Early dental evaluation in patients with WBS is important to prevent the development of carious lesions and endodontic infections and to identify different abnormalities that may be seen in cases.

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Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Mesude Çitir;* ***Design:*** *Mesude Çitir;* ***Control/Supervision:*** *Ayşe Zeynep Zengin;* ***Data Collection and/or Processing:*** *Mesude Çitir;* ***Analysis and/or Interpretation:*** *Ayşe Zeynep Zengin;* ***Literature Review:*** *Mesude Çitir;* ***Writing the Article:*** *Mesude Çitir;* ***Critical Review:*** *Ayşe Zeynep Zengin;* ***References and Fundings:*** *Mesude Çitir;* ***Materials:*** *Mesude Çitir.*

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