Case Reports and Reviews

Case Report

Diagnosis and Treatment of Gingival Enlargement in a Patient with Williams Syndrome: Case Report

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Abstract

Introduction: *Williams-Beuren syndrome (WS) is a rare genetic disease that affects approximately 1,20,000 live births. Patients with WS have specific skeletal deformities, distinctive facial features, growth retardation, mild to moderate intellectual disability, structural cardiovascular anomalies, abnormalities in dental morphology such as: peg-shaped incisors, bud-shaped maxillary first molars, microdontia, dental fusion and enamel hypoplasia, they also present class II and III malocclusion, anterior open bite, deep bite, anterior crossbite, tongue thrust, excessive interdental spaces and gingival enlargement.* ***Case presentation:*** *A 17-year-old male patient presented to the Periodontics Specialty of the Dental Hospital of the Catholic University of Honduras. Obtaining medical history revealed that the patient was diagnosed with WS immediately after birth. Upon questioning, the patient reported congenital heart disease; aortic stenosis and arterial hypertension.* ***Conclusions:*** *Due to the multiple manifestations in patients with Williams syndrome, it is important to make a thorough diagnosis and multidisciplinary treatment.*

Keywords

Gingival hyperplasia; Williams syndrome; Congenital Anomalies; Gingivectomy

Introduction

Williams-Beuren syndrome (WS), also known as “elf face” syndrome, was first described by Dr. Williams and Dr. Beuren in 1961-1962. It is a rare inherited genetic disorder that occurs in approximately 1,200,000 live births. It is caused by a hemizygous deletion in 26 to 28 contiguous genes, including elastin and LIM domain kinase 1 (LIMK1) in region q11.23 of chromosome 7 [1,2] Patients with SW are characterized by distinctive facial features, growth retardation, mild to moderate intellectual disability, gastrointestinal problems, dental abnormalities, structural cardiovascular abnormalities such as: supravalvular aortic stenosis (SAVS) (58%), ventricular septal defect (21%), mitral valve prolapse (15%) and aortic regurgitation (10%) and also non-structural abnormalities such as: arterial hypertension (40-50%) [3]. The diagnosis of SW is mainly based on the recognition of facial features, as well as on the frequency of EASV. However, phenotypic variability makes its diagnosis difficult, especially during the first months of life. Fluorescent in situ hybridization is the most widely used method for the molecular diagnosis of clinical SW [4,5]. In addition, multiplex ligation-dependent probe amplification, DNA markers, and array comparative genomic hybridization can be employed [6]. Numerous oral manifestations are described in patients with SW, including abnormalities in dental morphology such as: peg-shaped incisors, bud-shaped upper first molars, microdontia, dental fusion and enamel hypoplasia, Likewise, it presents class II and III malocclusion, anterior open bite, deep bite, anterior crossbite, lingual thrust, excessive interdental spaces and gingival enlargement [7]. In this study, the diagnosis and treatment of gingival enlargement in a patient with Williams syndrome is presented.

Case presentation

A 17-year-old male patient was presented to the Periodontology Specialty of the Dental Hospital of the Catholic University of Honduras referred from the Department of Dentistry of the Teaching Hospital for periodontal evaluation. Upon obtaining the medical history, it was revealed that the patient was diagnosed with SW immediately after birth, being the only case present in his family.

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Figure 1. *Characteristic facial clinical features (elf-like face) of SW including broad forehead, depressed nasal bridge, wide nose, and prominent open mouth. A. Frontal photograph. B. Lateral view. C, D. Facial symmetries.*

Upon questioning, the patient reported congenital heart disease; aortic stenosis and arterial hypertension that were diagnosed by ultrasound 4 days after birth. In addition, the patient has a mild intellectual disability with hypersocial behavior.

A set of extraoral photographs were taken to describe the facial characteristics. It was observed that the patient had a triangular face with a dolichofacial facial biotype, bipupillary dystopia, a wide forehead with asymmetric fifth facial features, a small straight nose with a wide base, large lips with a protrusive lip contour, a concave profile, and maxillary and mandibular retrognathism. (Figure 1)

Intraoral examination

The morphology of each tooth, position, interdental spaces, anomaly of number, shape of its arches and occlusion were recorded, dental caries is evaluated by means of the CPOD index which revealed the presence of 26 permanent dental organs of which the presence of caries was observed in relation to teeth 16, 12, 11, 21, 26, 36, hypomineralization of the enamel in the upper teeth, proclination of the upper and lower anterior teeth, presence of diastema, anterior open bite and lingual thrust habit was also perceived (Figure 2).

Periodontal examination

It was performed by two periodontists and measurements were taken using a UNC15 periodontal probe in which probing depth (PPD; distance measured from the gingival margin to the bottom of the gingival sulcus), clinical attachment level (CAL; distance from the cementoenamel junction to the bottom of the gingival sulcus) and bleeding on probing (BOP; percentage of

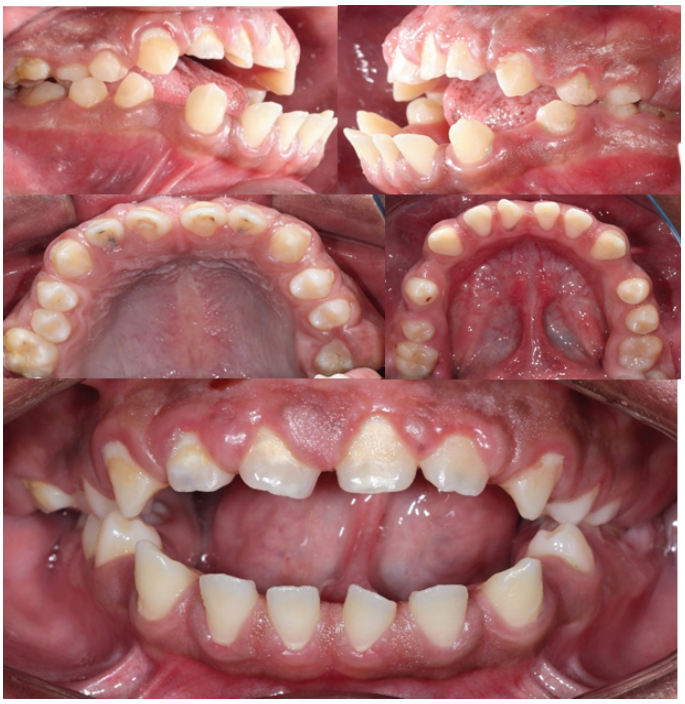

Figure 2. *Intraoral photographs*

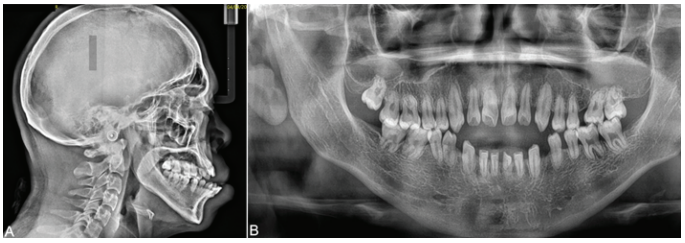

Figure 3. *(A) lateral skull. (B). Panoramic radiograph*

sites that bleed on probing in six areas for each tooth) were determined and the O'Leary index was taken to record plaque control. The patient presented gingival enlargement, bleeding on probing (30%) with an O'Leary index (80%). The periodontal phenotype was evaluated taking into account the thickness of the vestibular plate by using digital tomography, gingival thickness by the probe translucency method and the width of keratinized gingiva. The result of gingival thickness and bone width greater than 1 mm (thick flat phenotype).

Radiographic examination

A panoramic and lateral skull radiograph was ordered to perform Ricketts, Steiner and Jarabak cephalometric analyses in order to determine alterations in the position and size of the maxillae and their relationship with the teeth and other craniofacial structures. The panoramic radiograph showed asymmetrical condyles, a poorly permeable left nasal cavity, pneumatized maxillary sinuses, open gonial angles, anterior open bite and short dental roots. The lateral skull radiograph showed the vertebrae in lordosis, hyoid bone at C4 level, mandibular posterior rotation and compressed airway. (Figure 3)

Cephalometric Analysis

In the results of the Ricketts, Jarabak and Steiner analyses, the patient presents a dolichofacial biotype, vertical growth pattern, class III molar relationship, biprotrusion, incisor proclination, skeletal open bite, mandibular retrognathia, maxillary retrusion, short cranial base, labial protrusion and lower lip eversion.

Therefore, a diagnosis was made of vertical growth pattern, asymmetric facial thirds and fifths, concave profile, lip incompetence, muscular hypertonicity of the chin, class III molar relationship, short cranial base, and skeletal open bite.

Surgical Procedure

After obtaining approval of the treatment plan and informed consent, phase I of periodontal therapy was performed, which included removal of plaque and calculus supra and subgingivally from the upper and lower jaw. Prior to performing any treatment, antibiotic prophylaxis was indicated according to the American Heart Association guidelines for patients with congenital heart disease; which consists of oral administration of 2 g of amoxicillin one hour before the procedure. Instructions were given for performing oral hygiene using an electric toothbrush to facilitate hygiene by the patient.

One week later, phase II of periodontal therapy was started under 0.2% local anesthesia (lidocaine with epinephrine 1:100,000) using an infiltration technique in the vestibular buccal mucosa from the upper right first molar to the upper left first molar; measurements of the pseudopockets were taken with a UNC15 probe and gingivectomy was performed using an electrocautery. Due to the patient's limited openness and to avoid overly long appointments, gingivectomy of the mandibular area was performed in a second appointment (Figure 4). Postoperative pain was managed with 1 g of acetaminophen three times a day for three days. The patient was instructed to avoid hot drinks and foods for the first 24 hours and a 0.12% chlorhexidine gluconate mouthwash was recommended twice a day for two weeks. Follow-up appointments revealed exposure of the anatomical crowns of the teeth, improving aesthetics and allowing access to the caries in the posterior teeth for future restorations.

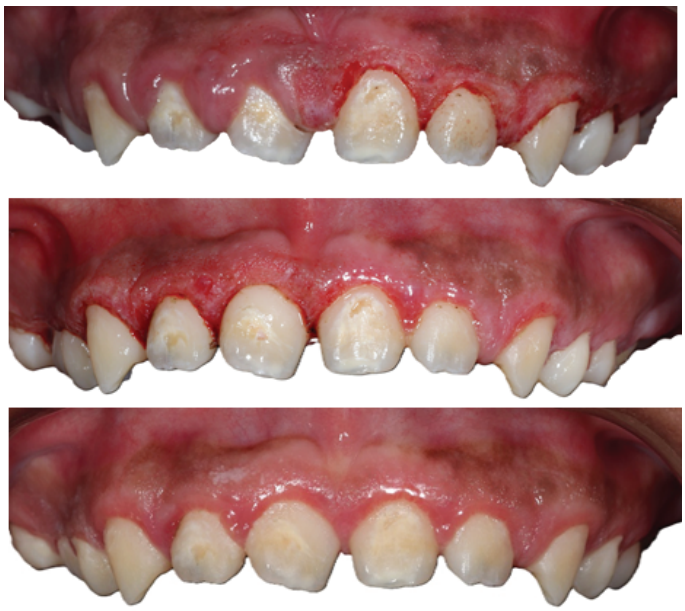

Figure 4. *(A, B).Gingivectomy and gingivoplasty. (C). Postoperative control 6 months later*

Discussion

In general, many of our dental findings are similar to the studies [8,9] where abnormal dental morphology, reduced mesio-distal crown dimension, excessive interdental spaces and enamel hypoplasia were observed.The genes Gtf2i, Gtf2ird1 and Gtf2ird2 are expressed during odontogenesis and are members of the TFII-I family that play a role in regulating the shape of the dental crown [10].

Regarding the radiological and cephalometric findings, patients with SW may present dental malocclusion, open bite, atypical swallowing, counterclockwise rotation of the maxilla and mandibular retrusion, as in this case, it presents a class III malocclusion and skeletal open bite, a fact that is related to what was reported by Scallop G et al. [11]. Periodontal phenotype classification was performed by measuring the thickness of the vestibular plate and by the translucency of the contrast probe, according to the method of Vavetsi et al. in which they classified the phenotype by palpation of the bone tissue and visualization of the gingival thickness [12]. To date, there are few studies that have examined the oral manifestations of SW and to our knowledge there are no studies that address gingival enlargement and accurately classify the periodontal phenotype using CBCT. In the present study, we describe the oral manifestations and surgical treatment of gingival enlargement in a patient with SW.

Conclusion

Due to the multiple manifestations in patients with Williams Syndrome, it is important to make a meticulous diagnosis in order to offer the best multidisciplinary treatment.

Conflict of interest

The authors report no conflict of interest.

Authors' contribution

Vilma Alejandra Umanzor: conceptualization, initial article review

Guillermo de Vicente: Research, bibliographic review, writing, initial draft, final draft

Katherine Santely: case diagnosis, cephalometric analysis, formal analysis

Linsey Peraza: Research, literature review, periodontal phenotype analysis

Hugo Romero: Validation, Initial Draft Writing

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