Dental management of patient with Williams Syndrome - A case report

Daniel Wong, Srinivas Sulugodu Ramachandra, Ashish Kumar Singh

Abstract

Williams syndrome is a multisystemic rare genetic disorder caused by deletion of 26–28 genes in the long arm of chromosome 7. It is characterized by developmental and physical abnormalities including congenital cardiovascular abnormalities, mental retardation, neurological features, growth deficiency, genitourinary manifestations, gastrointestinal problems, musculoskeletal problems, unique behavioral characteristics, and dental problems. Dental abnormalities include malocclusion, hypodontia, malformed teeth, taurodontism, pulp stones, increased space between teeth, enamel hypoplasia, and high prevalence of dental caries. Authors report a 17-year-old female patient with underlying Williams syndrome. Oral features and problems seen in the patient are listed. Malocclusion and screwdriver shaped teeth were noticed. Generalized widening of the periodontal ligament space with vital teeth was seen. This finding has not been reported in cases of Williams syndrome earlier. Precautions taken during dental treatment in patients with Williams syndrome are also discussed.

Keywords: caries, dental findings, periodontal disease, Williams syndrome

Introduction

Williams syndrome is a multisystemic rare genomic disorder that was first described, in 1961, by a cardiologist from New Zealand, Dr. J.C.P. Williams.[1] This syndrome is also called as Williams-Beuren syndrome after Dr. A J Beuren from Germany.[2] Williams syndrome is caused by a deletion of 26–28 genes in the long arm of chromosome 7 at 7q11.23. The deletion occurs at the time of conception. Williams syndrome has a prevalence rate of 1 in 10,000 individuals[3] to 1 in 25,000 individuals in certain ethnicities.[4] It is characterized by a number of developmental and physical abnormalities including congenital cardiovascular abnormalities, mental retardation, neurological features, growth deficiency, genitourinary manifestations, gastrointestinal problems, musculoskeletal problems, unique behavioral characteristics, and dental problems. Williams syndrome is usually diagnosed by fluorescent *in situ* hybridization (FISH) test, multiplex ligation-dependent probe amplification, or chromosome microarray, which detects the deletion of elastin gene on the long arm of chromosome 7.[4]

Studies have reported increased frequency of dental abnormalities in Williams syndrome, including malocclusion, hypodontia, malformed teeth, taurodontism, pulp stones, increased space between teeth, enamel hypoplasia, and high prevalence of dental caries. Because of these anomalies, the dentist contributes significantly to the successful overall management of these patients.[5] The presence of underlying medical disorders such as cardiovascular problems, hypertension, hypercalcemia, and kidney aliments may necessitate certain changes in the treatment protocol to be followed by the dentist.[6] In spite of the major advances in the diagnosis and possible supportive care for patients with Williams syndrome, the risk of sudden death still exists.[7] The risk of sudden cardiac death is 25–100 times greater in patients with Williams syndrome compared to the general population. The dental clinician should be knowledgeable of the precautions and protocol to be followed while treating patients with Williams syndrome. Authors report the dental features and conditions in a 17-year-old female patient with Williams syndrome.

Case Report

A female patient aged 17 years reported to Faculty of Dentistry, SEGI University for a routine dental check-up. The patient was of Chinese ethnicity and was accompanied by her mother. Patient's medical history revealed underlying Williams syndrome. Detailed history, extra-oral examination, intra-oral examination, and radiographic evaluations were performed. Extra-oral examination revealed the long face, broad forehead, wide cheeks, underdeveloped mandible, pouting lips, and widely spaced eyes. The characteristic "elfin facies" seen in Williams syndrome was noticed [Figure 1a]. Intra-oral examination revealed Angle's class III

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malocclusion [Figure 1b]. The mandibular right lateral incisor was screwdriver shaped [Figure 1b and c]. Plaque and calculus deposits, especially in the mandibular anterior region, deep pits and fissures and dental caries, was noticed in maxillary left and right second molars [Figure 1d]. Intra-oral periapical radiographs were taken which showed generalized widening of the periodontal ligament space [Figure 2a-d]. Pulp vitality tests revealed that the teeth were vital. No history of trauma was reported. A treatment plan involving oral prophylaxis, pit and fissure sealants, and restorations for carious teeth were planned for the patient. Due to the various systemic complications associated with Williams syndrome, the patient was referred to the physician for medical opinion and consent for the above mentioned dental treatment.

Patient's physician provided the medical consent for dental treatment, along with the medical history of the patient. Williams syndrome was diagnosed by FISH test 7q11.2 deletion. The patient had a history of epilepsy for which she was taking anticonvulsants. However, anti-convulsants were stopped since 2 years in the patient. Ultrasound examination of the kidney and the neural artery Doppler studies were also normal in the patient. The patient had a history of transient hypertension, but her blood pressure was normal without any medication. The patient had some learning difficulties but was otherwise very independent, cheerful and friendly with good self-esteem.

The treatment needed for the patient was explained, and informed consent was obtained from the patient. Oral prophylaxis, composite restorations in maxillary right and left second molars, and pit and fissure sealant in mandibular right and left first molars were performed in the patient. The patient was advised for regular follow-up and an orthodontic consultation for malocclusion.

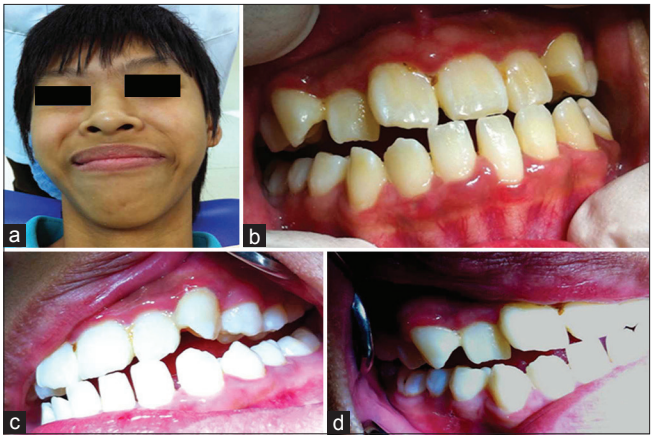

Figure 1: (a) Extra-oral image of a 17-year-old female patient with Williams syndrome. The characteristic "elfin facies" seen with a long face, broad forehead, wide cheeks, underdeveloped mandible, pouting lips, and widely spaced eyes can be appreciated. (b) Clinical image is showing anterior teeth. The mandibular right lateral incisor, which is screwdriver shaped can be noticed. (c) Clinical image of left posterior teeth. The healthy condition of the gingiva is to be noted. (d) Clinical image of right posterior teeth. Carious buccal pit on the mandibular right first molar can be noted

Discussion

Patients with Williams syndrome are social and friendly in nature. Extreme interest toward music has been noticed in these individuals. Our patient was socially active and cheerful in nature. In patient's with Williams syndrome, initial medical evaluation (especially of the cardiovascular system, renal and nervous system) is important. Aging results in increased aortic stenosis and progressive renal impairment. These complications result in increased blood pressure. In our patient, the medical evaluation revealed that her blood pressure was normal despite the patient not being on anti-hypertensive medications. Her last episode of epilepsy was 4 years back with anti-epileptics being stopped 2 years back.

Precautions during dental treatment for patients with Williams syndrome

A medical evaluation to rule out cardiovascular abnormalities and renal abnormalities, which could affect dental treatment is mandatory. Cases of sudden death have been reported with administration of anesthetic in these individuals. Hence, caution should be exercised while administering anesthesia in these individuals.[7] Individuals with Williams syndrome are hypersensitive to certain frequencies of sound (hyperacusis).[8] Hence, any form of dental equipments such as ultrasonic scalers, highspeed handpieces, which generate sound should be first demonstrated, before being used on the patient.

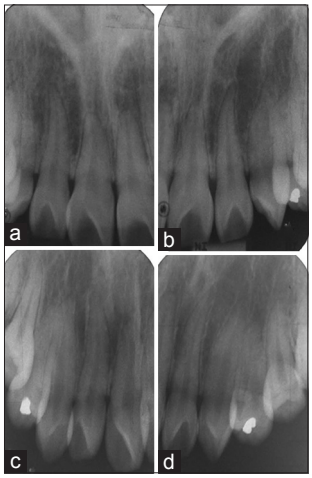

Figure 2: (a) Intra-oral periapical radiograph showing the right maxillary incisors. (b) Intra-oral periapical radiograph is showing the left maxillary incisors. (c) Intra-oral periapical radiograph is showing the right maxillary premolars. (d) Intra-oral periapical radiograph is showing the left maxillary premolars. Widened periodontal ligament space around the roots of the teeth can be appreciated

Intra-orally, malocclusion has been one of the commonly reported findings.[5] Our patient had Angle's class III malocclusion. Screwdriver shaped teeth are the other commonly reported dental finding,[5] which was also noticed in our case. Generalized widening of the periodontal ligament space was noted, which has not been reported in earlier cases of Williams syndrome. Pulp vitality tests revealed that the teeth were vital. Periodontal evaluation of the patient revealed a healthy periodontium. Patients with Williams syndrome have normal periodontium and are not predisposed to increased periodontal destruction, despite the presence of elastin gene haploinsufficiency.[9] Patients with Williams syndrome have increased chances of impaired glucose tolerance with reduced insulin sensitivity.[10] These are traditional risk factors for type 2 diabetes mellitus. Hence, the patient was advised to undergo estimation of glycemic levels periodically.

There is neither a cure for Williams syndrome nor a standard treatment protocol. Health care for these patients is mainly symptomatic and supportive. Individuals with Williams syndrome need regular monitoring for potential medical and dental problems by health care professionals who are familiar with the disorder. The prognosis for individuals with Williams syndrome varies. Some may be able to master self-help skills, complete academic, or vocational school, and live in supervised homes or on their own while others may not progress to this level. Apart from this there are numerous associations for individuals with Williams syndrome, which provide resources, support, and latest medical information on the syndrome to all concerned (patients, caregivers/families, and medical professionals).[11]

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