Comprehensive Dental Rehabilitation of a Patient with Williams-Beuren Syndrome - A Case Report

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ABSTRACT

Williams-Beuren Syndrome (WBS) is a rare congenital disorder inherited on an autosomal dominant pattern. These patients often present with dysmorphic craniofacial features in addition to supravalvular aortic stenosis and cognitive impairment affecting their wellbeing. Unfortunately, the literature is sparse on the dental management of these patients. This case reports describes a patient with WBS who presented with the associated dento-facial characteristics. This report also outlines the interdiscliplinary dental management of the patient through combined orthodontic–orthognathic surgery – restorative procedures.

KEYWORDS: Orthognathic Surgery, William's Syndrome, Dental Rehabilitation

INTRODUCTION

Williams Syndrome is a rare congenital disorder resulting from a genetic alteration in the long arm of chromosome 7. There is hemizygous deletion of the elastic gene on chromosome 7q11.23[1]. Also known as Williams-Beuren Syndrome (WBS), these patients typically present with dysmorphic craniofacial features, supravalvular aortic stenosis and cognitive impairment that is associated with an autosomal dominant hereditary inheritance pattern. This poses a 50 % probability of transmission of this disorder to the offspring. The prevalence of WBS can vary to up to 1:20,000 live births[2]. However, according to the Williams Syndrome Association, the prevalence is about 1:10,000 live births[3]. It has also been reported that it might be even greater as many go undiagnosed. In addition, there is no distinction of prevalence between sex, race, and ethnicity[4].

The clinical manifestations are multi-systemic and can affect the cardiovascular, neurological, renal, visual, and auditory systems[5]. Most affected individuals have congenital heart defects, most often supravalvular aortic stenosis and peripheral pulmonary stenosis. Other very common signs include delayed and impaired cognitive development, poor motor coordination, irritability, hyperactivity and anxiety. There are also records of renal dysfunction, hypertension, hypercalcemia, and reduced visual and auditory acuity[6].

The facial appearance often is said to be "elfin" like, is characterised by a broad forehead, strabismus, prominent ears, small nose with wide tip, depressed nasal bridge, flattened midface, long philtrum, increased inter-commissure distance, thick lips and full cheeks. They may present either with a Class II and III skeletal patterns often with increased maxillary-mandibular plane angles, anterior inclination of the maxilla and a deficient bony chin[7].

Dental features often include excessive interdental spacing, hypodontia, microdontia, enamel hypoplasia and macroglossia[8]. Literature is sparse regarding possible dental and orthodontic interventions in patients with WBS. The following is a case report of a patient diagnosed with WBS who received multidisciplinary dental care to restore occlusal function, aesthetics and general well –being of the patient.

CASE REPORT

We describe a case of a 25 year old male with WBS who was accompanied by his father, an armed forces veteran, whom presented to the Tuanku Mizan Dental Specialist Polyclinic, Kuala Lumpur, Malaysia requesting for orthodontic treatment to improve his bite.

Medical History

Diagosis of WBS was made by the patient's pediatrician when he was a child at University Kebangsaan Malaysia, Kuala Lumpur where the patient had been attending for all his medical care. Genetic testing by fluorescent in-situ hybridisation analysis (FISH) confirmed of the WBS diagnosis together with the clinical signs that he presented with when he was a child. Generally, the patient was well although he has mild aortic stenosis that did not pose any functional problems for him thus far. He did not have hypertension. He has hypercalceamia that was well controlled by medication Calcitonin. Though intellectually compromised, the patient was very well motivated and was able to communicate his concerns well to the dental team on a whole. He has a small physique, short stature standing at just 5 feet tall. He suffers with strabismus, reduced visual acuity, hyperacusis and a hoarse voice. This patient reported very minimal experience with dental treatment as a child.

Extra-oral Features

The patient presented and displayed a Class III skeletal pattern with increased Frankfort Mandibular Plane Angle (FMPA) (Figure 1). The steinocleidomastoid muscles are very prominent and there is right sided torticollis. The lower lip is thick and prominent, and the lips were competent at rest. There is no vertical maxillary excess. The philtrum length is long at 25mm. There was no major facial asymmetry noted.

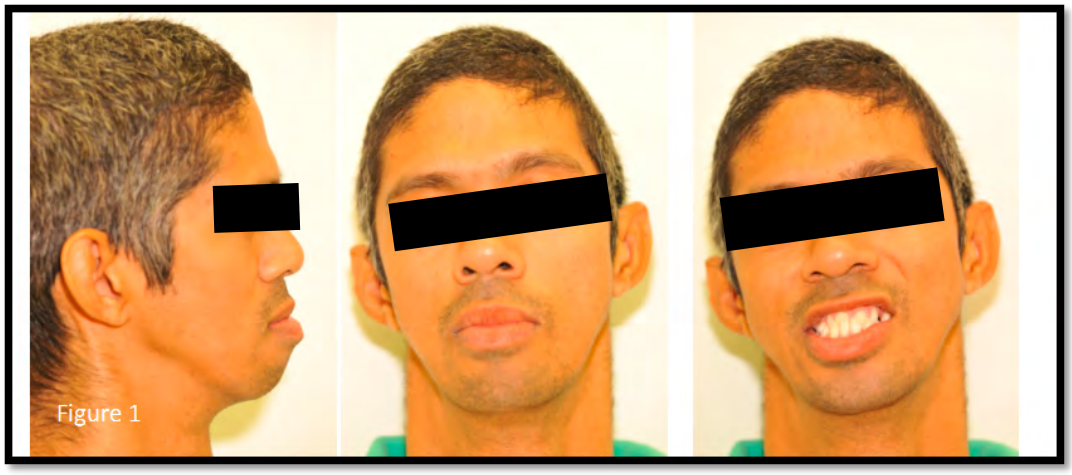


Figure 1: Initial Photos - Extraoral views

Intra-Oral Features

Soft tissues examination reveals macroglossia. Basic periodontal examination confirmed periodontal attachment loss around teeth 15, 25, 24, 22, 34, 33, 43 and 44 although there were no periodontal probing depths beyond 2mm. There is oligodontia, a condition where there is congenital absence of six or more teeth, excluding third molars and generalised microdontia (small sized teeth). The clinically missing teeth are 18, 16, 13, 23, 26, 28, 38, 36, 35, 32, 42, 46 & 48. The 14 and 15 were severely rotated mesio-palatally. The occlusion is in complete anterior and posterior crossbite with reverse overjet at 3mm with no associated displacement.The overbite is incresed at 80%.The dental centrelines were coincidental. His oral hygiene was fair with no evidence of dental caries.

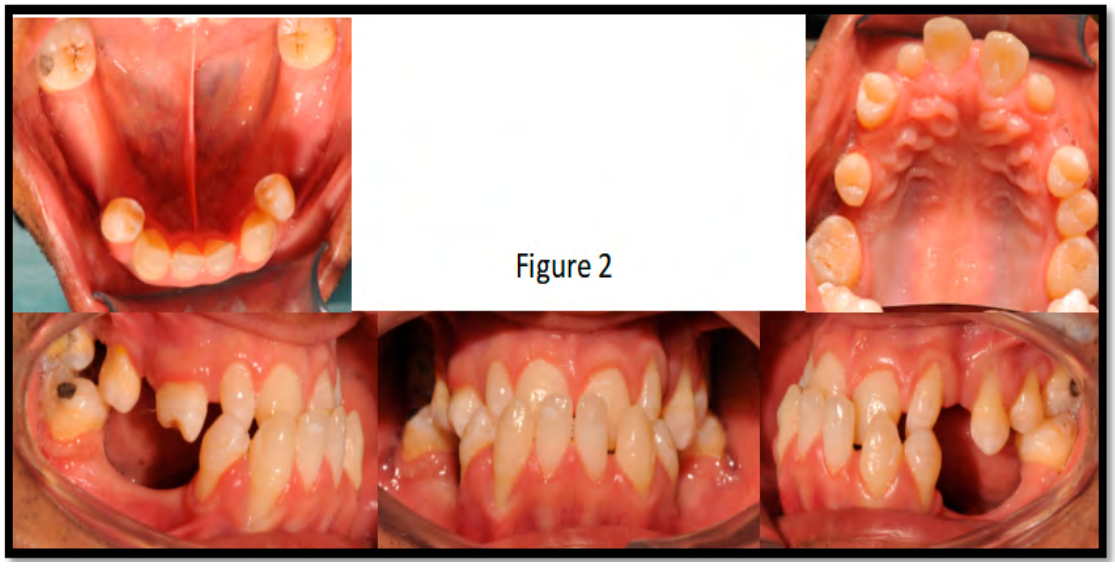


Figure 2: Pre Treatment - Intraoral views

Dental panoramic tomogram (DPT) (Figure 3) confirmed the missing teeth and presence of ectopic lower right premolar (45) being horizontally impacted at the right angle of the mandible close to its inferior cortical border. The angles of the mandible appeared slender bilaterally.

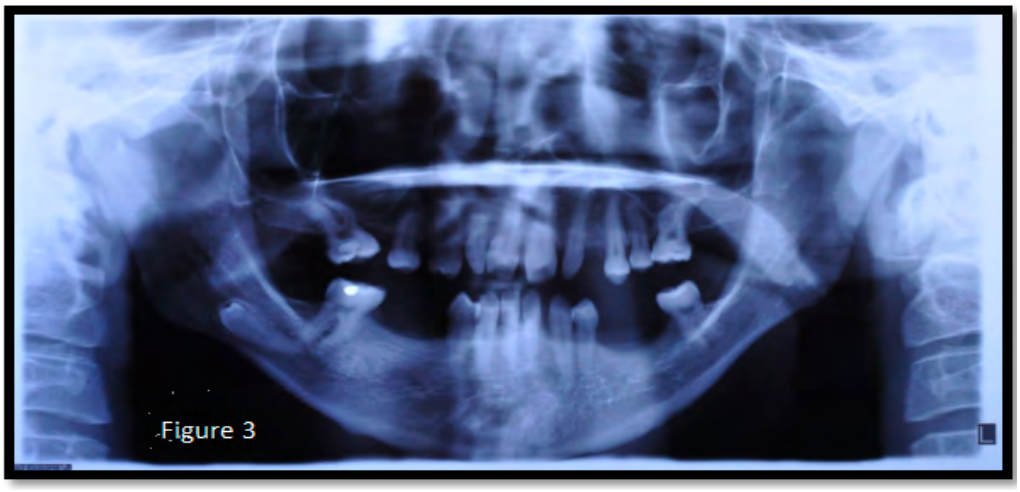


Figure 3: Pre Treatment - DPT

Treatment Plan

At the outset, several options were discussed in view of the patient's condition. The initial treatment plan was to keep orthodontic treatment simple by space redistribution to facilitate dental rehabilitation by partial dentures, accepting the Class III incisor relationship. However, the patient was adamant on comprehensive orthodontic/orthognathic treatment for his dental condition. Upon obtaining medical and anaesthetic clearance for the patient to undergo orthognathic procedure, the following treatment plan was agreed upon with informed consent obtained:

1. Secure optimum oral hygiene
2. Straight Wire Appliance (Fixed Appliance MBT™ 022-inch slot bracket)
   a. Level and align the occlusion
   b. De-rotate the upper right premolars (14 &15)
   c. Approximate the anterior spaces of the labial segment.
3. Decompensate and coordinate the arches for orthognathic surgery.
4. Surgical planning to achieve a Class I incisor relationship.
   a. Maxillary advancement 4mm with 2mm posterior impaction.
   b. Minimal clockwise autorotation of the mandible
5. Post-op settling of the occlusion.
6. Hawley retainers with pontics.
7. Composite build-up of the dimunitive upper right & left lateral incisors(12 & 22).
8. Cobalt chromium partial dentures of both arches.
9. Review.

Treatment

Fixed appliance was commenced using straight wire edgewise technique with MBT™ prescription 0.022-inch slot bracket system upon establishing an optimal oral hygiene routine. A mini-bracket system (Mini Masters® American Orthodontics) with a smaller base and diagonal angulation torque system was used as the teeth surfaces were small. Round and small diameter Nickel Titanium archwires were used to derotate the rotated premolars and gain initial levelling and aligment. Complete levelling and alignment of the mandibular and maxillary arches-,dental decompensation,space redistribution and harmonising the upper and lower arches were carried out in posted 0.019 x 0.025 inch Stainless Steel archwires. The anterior teeth were approximated using elastomeric power chains. All remaining spaces posteriorly were consolidated at the edentulous areas anterior to the last standing molars. This pre-surgical orthodontic phase took 14 months to achieve (Figure 4).

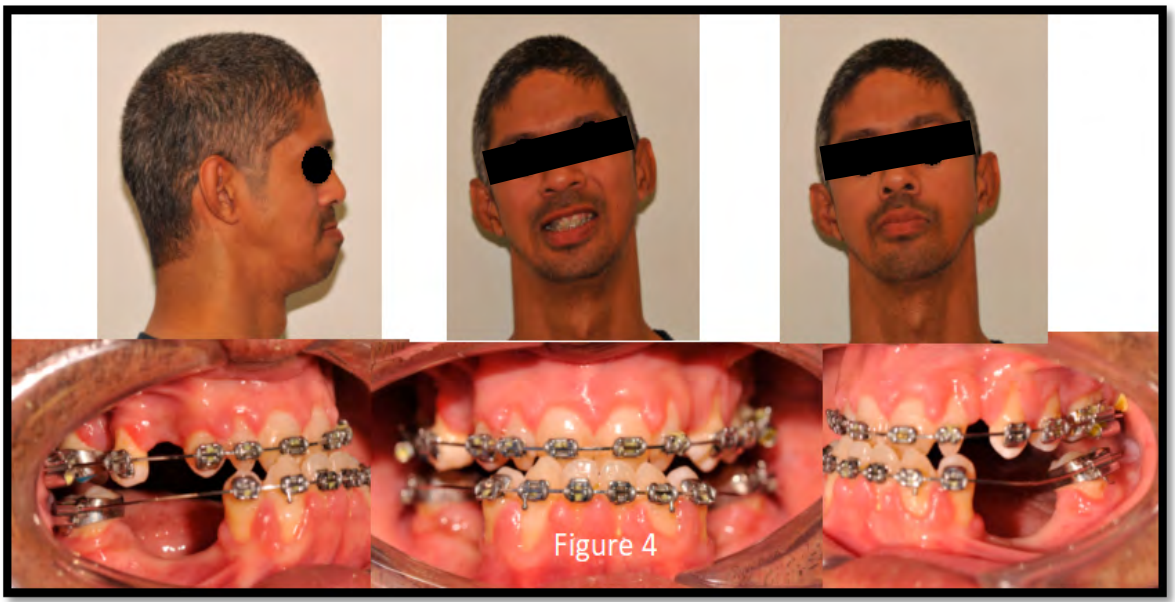
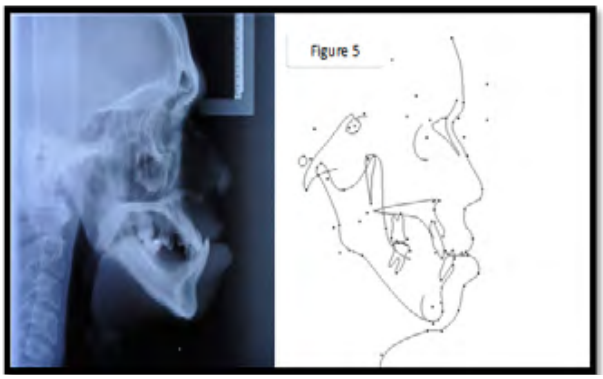
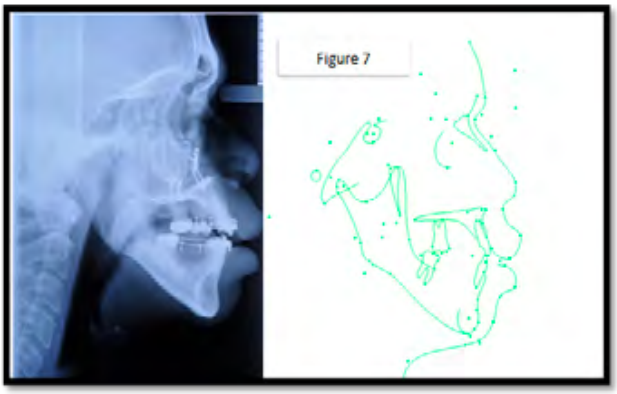


Figure 4: End of Pre-surgical Orthodontic Treatment.





Figures 5,6 & 7: Lateral Cephalograms and Cephalometric Assessments at the Start of Treatment, Pre- Orthognathic Surgery and Post-Surgery Respectively.

Lateral Cephalometric data analysis in Table 1 confirmed that the patient presented with a Class III skeletal pattern with a hypoplastic maxilla, prognathic mandible, an increased gonial angle and maxillary mandibular (MM) angle. Despite the high MM angle, the antegonial notching was not very pronounced. The lower incisors were upright for the given MM angle though the upper incisors were proclined.

Outcome of treatment:

Figure 8 shows the occlusion at the end of orthodontic treatment where there was a Class I incisor relationship with a stable bilateral crossbite relationship on the last standing molars. The occlusal plane had been restored with a stable overjet and overbite despite a 1mm lower dental centreline shift to the right hand side. The 12 & 22 have been restored. The patient was issued with a upper and lower Hawley retainers with pontics at the edentulous region whilst waiting for his tooth-borne cobalt chrome denture to be constructed and fitted.

Table 1: Lateral Cephalometric Analysis (Eastman Analysis)

Eastman Analysis	Normal	Initial	Post-treatment
SNA (°)	81	77	85
SNB (°)	78	83	82
ANB (°)	2	-7	3
SNPog Angle(°)	80	84	82
Maxillary-Mandibular angle(°)	27	45	40
Upper facial height (UFH) (mm)	47 ± 2	40	39
Ratio LFH/Total Face Height (%)	57.3 ± 2	59	57
Angle U1 to Palatal plane(°)	109.6 ± 6	113	104
Distance lower incisors to A-Pogonion line (mm)	1.0 ± 2	12	2
Angle of lower incisors to Mandibular plane(°)	92	77	74

Figure 8: Final Occlusion After Fixed Appliance Removal.

Figure 9: The Final Occlusion with the Cobalt Chrome Denture in Place.

Figure 10 : Cephalometric Superimpositions

Orthognathic surgical treatment was carried out by the Maxillofacial Surgical team by advancing the maxilla by 5mm and posterior impaction of 3 mm using Le Fort 1 technique with a minimal clockwise autorotation of the mandible to achieve Class I incisor relationship. A stable occlusal contact on the last standing molars of each quadrant was established so as not to lose the planned occlusal vertical dimension of the patient and risk overclosure of the mandible given that the patient is only left with one madibular molar bilaterally. The maxilla was secured with angled miniplates bilaterally. Post-surgery, no intermaxillary fixation was used though the occlusion was settled using orthodontic elastics. Three (3) months after his surgery, upon completion of his orthodontic treatment, the appliances were removed, and the patient was prescribed with Hawley retainers with pontics to restore the edentulous

DISCUSSION

Adult cranio-facial syndromic patients who present to us are rare in the Dental Services of the Malaysian Armed Forces. For those who do present to us, it poses many challenges for the team to provide comprehensive dental treatment. The syndromal, skeletal and dental manifestations or malformations often require individualised and complex treatment planning to manage the patient holistically. Full mouth rehabilitation provided for this patient involved collaboration between the dental specialties over a duration of 3 years inclusive of preparatory work, orthodontics, orthognathic surgery and finally with prosthodontic care using cobalt-chrome partial dentures. This patient, although suffering from WBS showed exemplary co-operation and compliance which was vital towards the success of his treatment. In patients who are less compliant, a lesser treatment objectives by accepting the skeletal and dental framework of the patient would have been preferred.

The Cephalometric superimpositions of pre-treatment(black) & post-treatment(green) show that the maxilla has been advanced with posterior impaction as planned. The vertical dimension of the patient largely remained the same. From Figure 10 and the post-treatment values in Table 1, the lower molars were protracted and the lower incisors where somewhat over-retracted with during the lower anterior space closure and space redistribution stage. It is well known that anchorage control and orthodontic tooth movement are difficult to predict and control using standard sized bracket for those with microdontia.

In Asian populations, prevalence of hypodontia has been reported up to 6.4%9. Syndromic patients such often present with dental malformations. In this case, the patient had generalised microdontia and oligodontia. It is well known that hypodontia may occur either as part of a syndrome or as a non-syndromic form. However, nonsyndromic hypodontia is more common, with varying numbers of teeth that can be involved10. Biomechanics of treating patients with many missing teeth and small teeth can be particularly complicated. Where possible, it is best to limit the tooth movement planned.

With small teeth come the associated thin roots which are at higher risks of root resorption when subjected to orthodontic forces11. It was imperative to use gentle and interrupted forces to orthodontically move the teeth to avoid external apical root resorption. Duration between the appointment for archwire changes and space redistribution were deliberately prolonged to 8 weeks intervals instead of the 6 weeks to allow ample time for repair cycle of the root surfaces to take place during the resorption-repair stages of orthodontic tooth movement[12].

There are numerous studies that suggest that partial glossectomy is recommended to improve stability of Class III orthognathic cases which present with macroglossia[13]. This was avoided due to the potential risks and complications that might occur including, excessive bleeding, decreased movement of the tongue, residual speech amd masticatory problems and anaesthesia of the tip of the tongue. Since the team had planned for a Le Fort 1 maxillary advancement surgery with posterior impaction, the space for the tongue was largely unchanged as the position of the mandible was only minimally altered post-surgery. The ectopic 45 was left undisturbed as there was no clinical indication for removal. However, review of the patient every 5 years to examine for cystic enlargement or infection was suggested.

The Le Fort 1 Osteotomy is a common procedure used by maxillofacial surgeons to correct a wide range of dentofacial deformities. It allows for correction in three dimensions including advancement, retrusion, elongation, and shortening of the maxilla. The Le Fort 1 osteotomy is named after the fracture pattern

originally described by Rene Le Fort in 1901 that extends from the nasal septum, along the tooth apices, and through the pterygomaxillary junction. Although the first description of a Le Fort 1 surgery by Cheever in 1864 was mainly described for resection of nasopharyngeal tumors, recent studies have focused on the reliability of maxillary movements as an orthognathic intervention with reliable long-term results[14].

The post-surgical appearance of the patient improved his self-confidence. He has obtained a part-time job at a local grocer and according to his parents, socialising with a larger circle of friends. With improved diagnostic and medical care available for patients with WBS, many of them have assimilated well into society. It is important for the dental team to recognise the characteristic signs and symptoms of those with Williams syndrome to better understand and manage their dental conditions comprehensively.

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