



Ashish Shah

Sunil Hirani and Jeremy Hodgkins

Orthodontic Management of a Patient with Williams Syndrome

Abstract: Williams Syndrome is a rare genetic condition with numerous systemic and dental manifestations. This case report highlights the orthodontic management of a 13-year-old female with Williams Syndrome, who presented with hypodontia and other dental anomalies.

Clinical Relevance: Dental practitioners should be aware of the factors affecting the dental and orthodontic management of patients with syndromes.

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Williams Syndrome (WS) is a multi-system congenital disorder characterized by numerous developmental and physical abnormalities. These include cardiovascular anomalies (congenital heart defects affect 53–80% of patients with WS), intellectual and developmental disabilities (in 75–95% of patients with WS), microcephaly (found in 65% of individuals with WS), reduced perceptual and motor functions, craniofacial features, an unusual elfin facies, pre- and post-natal growth deficiencies, genito-urinary, gastro-intestinal and musculoskeletal problems, impaired renal function, micrognathia and many dental anomalies. Affected individuals are more likely to have attention deficit and hyperactivity disorder, and about 80% exhibit generalized anxiety disorder; 74–95% are sensitive to sounds.^{1,2,3}

Williams Syndrome was first described in 1961 as an association of supravalvular aortic stenosis and intellectual and developmental



Figure 1. Facial features of Williams Syndrome.

disabilities in four children with characteristic facies.⁴ It is estimated to affect between 1 in 7500 and 1 in 50000 individuals.^{1,5,6} It is thought to be caused

by a microdeletion in the long arm of chromosome 7.¹ The microdeletion of the WS critical region on one of the two chromosomes 7 is detectable in 90–97% of individuals with the clinical phenotype of WS.⁵ Both sexes are affected equally.^{5,6} It can occur in all ethnic groups.^{5,6}

Cardiovascular and renal impairments are the main problems complicating dental treatment for children with WS. These tend to progress with age, so the severity of the disease should be determined prior to any dental treatment.¹

Short stature and premature puberty with an early growth spurt occur frequently in WS.⁵ This would definitely have an impact on the timing of orthodontic treatment. In addition to the systemic manifestations, orthodontists must pay particular attention to the dental and facial features of patients with WS.

This is a case report of a female patient with WS who presented with particular orthodontic challenges. To our knowledge, the orthodontic implications

Ashish Shah, BDS, MFDS RCS(Eng), Specialist Registrar in Orthodontics, Royal Berkshire Hospital (Reading), Wexham Park Hospital (Slough) and Guy's, King's and St Thomas' Dental Institute, **Sunil Hirani**, BDS, BSc, FDS RCS(Eng), MSc, MOrth, FDS(Orth), Consultant Orthodontist, Luton and Dunstable Hospital, Bedfordshire and **Jeremy Hodgkins**, BDS, FDS RCS(Eng), DOrth RCS(Eng), MScD, Consultant Orthodontist, Royal Bournemouth Hospital, Bournemouth, UK.



Figure 2. Anterior crossbite and mandibular arch spacing.



Figure 3. Maxillary arch crowding.



Figure 4. Screwdriver-shaped mandibular incisors.

and management of patients with WS have not been discussed before.

Description of case

A 13-year-old female (Figure 1) with WS and a repaired cleft palate was referred to the Orthodontic Department at the Royal Bournemouth Hospital for management of a Class III malocclusion. A diagnosis of WS

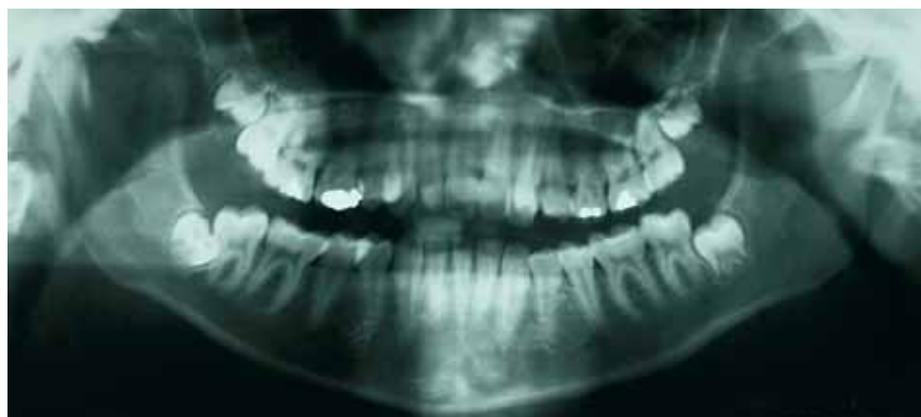


Figure 5. Pre-treatment dental panoramic tomography.

SKELETAL		
	Actual	Mean
SNA	81.5°	81.5°
SNB	80.5°	78.0°
ANB	1.0°	3.0°
SN/MxP	1.5°	7.5°
MxP/MnP	47.0°	27.0°
LAFH/TAFH%	50.5	56.0
TEETH		
	Actual	Mean
Overjet	-3.0 mm	3.5 mm
Overbite	4.0 mm	4.0 mm
UI/MxP	89.0°	109.0°
LI/MnP	75.5°	92.0°
LI-Apo	9.5 mm	1.0 mm

Table 1. Eastman Analysis of pre-treatment lateral cephalogram.

was confirmed by fluorescent *in situ* hybridization (FISH) when the patient was 12 years old. FISH is a cytogenetic technique that uses fluorescent probes to detect the presence of specific DNA sequences on chromosomes. The patient's cardiologist confirmed that she had a structurally normal heart and did not require antibiotic prophylaxis for dental treatment. To our knowledge, cleft palate in a patient with WS has only been reported once in the literature.⁷

Extra-oral assessment

- Skeletal Class III pattern;
- Reduced lower face height;
- Increased Frankfort mandibular planes angle;
- At rest, lips were competent;
- Acceptable facial profile;
- No mandibular asymmetry;

Intra-oral assessment

- Repaired cleft palate;
- Mild mandibular arch spacing (Figure 2);
- Severe maxillary arch crowding (Figure 3);

- Molar relationship Class III bilaterally;
- Teeth absent: UR5, UL5, LL2;
- Invaginated and diminutive UR2 (the invagination had been restored with composite resin by the patient's community dental officer);
- Screwdriver-shaped permanent mandibular incisors (Figure 4);
- Reverse overjet with a forward displacement of the mandible on closure into centric occlusion;
- Lower centreline shifted to the left side as a result of the developmentally absent LL2.

Radiographic assessment

Difficulties were encountered in taking ideal quality radiographic images as a result of the patient's limited motor skills. The pre-treatment dental panoramic tomograph (DPT) (Figure 5) and upper standard occlusal radiograph (Figure 6) showed the following anomalies:

- Hypodontia with missing UR5, UL5 and LL2 ;
- UR7 and UL7 distally angulated;
- Slender, pipette-shaped roots.

The DPT also revealed possible caries on the UL6. Dietary advice was given and the carious lesion investigated and restored by the patient's community dental officer.

A lateral cephalogram was taken (Figure 7). An Eastman Analysis (Table 1) confirmed the Class III skeletal relationship. A steep maxillary-mandibular planes angle was also noted.

Aetiology of the malocclusion

- **Skeletal:** Class III skeletal base;
- **Dento-alveolar:** developmental

absence of UR5, UL5 and LL2;

- **Soft tissues:** retroclined lower labial segment due to lower lip activity;
- **Habits:** nil.

Aims of treatment

- Preserve facial profile;
- Relieve crowding in maxillary arch, close spaces in mandibular arch;
- Attain Class I incisor and molar relationships;
- Obviate the need for prosthodontic treatment;
- Space closure in the maxillary arch would allow correction of the distally angulated UR7 and UL7.



Figure 6. Pre-treatment upper standard occlusal.



Figure 7. Pre-treatment lateral cephalogram.



Figure 8. The mandibular arch after orthodontic treatment.

Treatment plan and rationale

On discussion about the advantages and disadvantages of space opening and closure, the patient and her parents were keen to avoid prosthodontic treatment. Therefore, the following treatment plan was tailored to the patient’s needs:

- Extraction of URE and ULE;
- Insertion of Nance palatal arch to reinforce anchorage;
- Maxillary and mandibular fixed appliances with intermaxillary traction;

Treatment sequence

- Extract URE and ULE;
- Cement palatal arch;
- Lower removable appliance with incisor and posterior capping to unlock occlusion;
- Upper fixed appliance to align arch and improve incisor relationship;
- Lower fixed appliance to complete alignment.

Treatment results

Both arches were well aligned post-debond (Figures 8 and 9). The incisor and molar relationships were Class I and positive overjet and overbite were attained (Figure 10).

As the LL2 was developmentally absent, this meant that the maxillary and mandibular centrelines would not be coincident. However, the mandibular centreline was of minor aesthetic concern.

Discussion

Facial

The facial features of WS patients are distinctive and become more apparent with age. They include a flat midface, depressed nasal bridge, anteverted nostrils, long philtrum, full cheeks, thick lips, wide intercommissural distance, open mouth and low-set

ears. Ocular findings include medial eyebrow flaring, short palpebral fissures, hypotelorism, epicanthic folds, periorbital fullness and strabismus.^{2,8} Some of these characteristics can be seen in our patient (Figure 1).

The facial dysmorphology in WS is composed of soft tissue and skeletal components. Mass and Belostoky studied the skeletal component by examining the lateral cephalometric radiographs of eight children with WS.⁸ They concluded that four skeletal features may contribute to the characteristic facial appearance of WS:

- A short anterior cranial base, despite a normal cranial base angle;
- A steep-angled mandibular plane combined with a normal facial height;
- An unusual proportion of upper-to-lower anterior face height and posterior-to-anterior face height;
- A deficient chin button.⁸ However, the soft tissue characteristics may have a greater influence on the facial appearance.

In our case, the patient exhibited a steep-angled mandibular plane (47°) with a reduced lower facial height (50.5°). We suspect this is the result of the patient’s mandible being displaced forward into centric occlusion. This contributed to overclosure and therefore a reduced lower facial height.

Dental anomalies

The dental anomalies associated with WS patients include a mandible that is often small in contrast to a prominent maxilla.⁹ The maxillary arch has been described as being too broad for the mandibular arch.² However, these features were not found in our case. Hertzberg *et al*¹⁰ found 31.8% of individuals with WS to have a Class II malocclusion and 9.1% to have a Class III malocclusion.

Studies of patients with WS have reported increased frequencies of hypodontia, microdontia, invagination



Figure 9. The maxillary arch after orthodontic treatment.



Figure 10. The occlusion after orthodontic treatment.

of maxillary incisors, small and slender roots, pulp stones, excessive interdental spacing, malocclusion, delayed mineralization, enamel hypoplasia and mandibular second molar taurodontism.^{1,6,10} Hertzberg *et al*¹⁰ found that microdontia is an almost universal finding in WS. Axelsson *et al*⁶ found significantly smaller mesio-distal and labio-lingual dimensions for most tooth crowns in WS compared with the general population. Axelsson *et al*⁶ also found a higher prevalence of hypodontia (40.5%) of permanent teeth in WS than in the general population. They also found the frequency of agenesis of more than six teeth to be 11.9%, with a clear dominance of males (four out of the subgroup of five). The missing permanent teeth in the maxilla usually include second premolars, first premolars and lateral incisors. In the mandible, second premolars, first premolars and central incisors are the most commonly missing permanent teeth.⁶

Hertzberg *et al*¹⁰ recorded abnormal tooth morphology in 12.5% of the primary dentitions and 40.7% of the permanent dentitions in patients with WS. Axelsson *et al* found morphological aberrations of tooth crowns to be common in patients with WS. A high proportion of maxillary and mandibular incisors were described as tapered or screwdriver-shaped, with the exception of mandibular lateral incisors.⁶

Of the dental features associated with WS, our patient had hypodontia, invagination of the UR2, slender roots and screwdriver-shaped incisors.

Orthodontic considerations

Hypodontia and microdontia pose difficulties with orthodontic anchorage and mechanics. Small and slender roots are at greater risk of root resorption. Excessive interdental spacing may require a multidisciplinary approach involving restorative dentistry. Late mineralization would delay the onset of orthodontic treatment and enamel hypoplasia may cause failure of orthodontic bracket bonding. Most patients with hypodontia also tend to have microdontia. This has an effect on the 'golden proportions' of the upper labial segment and presents another aesthetic challenge.

A vast range of complications can occur in patients with hypodontia and each case should be considered individually and discussed fully within the multidisciplinary team.¹¹ However, several issues commonly arise in the orthodontic management of patients with hypodontia. These include:

- Space management;
- Uprighting and aligning teeth;
- Management of deep overbite;
- Retention and stability.¹¹

The decision on whether to open or close space depends upon the malocclusion, the age of the patient, the severity of hypodontia and the degree of inherent crowding.¹¹ Where space closure is possible, the need for a prosthesis, with its associated long-term maintenance, is avoided.¹¹ However, space closure in hypodontia cases may be slower than normal and it may not be possible to close spaces completely. Possible explanations include reduced alveolar bone mass, generally smaller teeth and anchor units that are weaker due to the smaller size and number of teeth.¹¹

In cases with moderate crowding, the crowding should be assessed as if all permanent teeth were present, and then developmentally absent teeth considered as 'extractions' to relieve the crowding.¹¹

Attempts to close substantial amounts of space tend to retract the anterior teeth.¹¹ This was undesirable in this case as the patient had a Class III malocclusion. A round stainless-steel archwire may be used to allow space closure with less friction. A rectangular archwire can be used if there is concern regarding loss of torque and excessive tipping of teeth.¹²

Early orthodontic assessment is recommended as simple intervention may minimize or prevent the development of malocclusions.

Conclusion

The ideal goals of orthodontic treatment (aesthetics, function and stability) are not always achievable and a compromise may have to be accepted.¹¹

A detailed clinical history, thorough diagnosis and liaison with specialists in a multidisciplinary environment can help to ensure optimal patient care. Treatment must

always be tailored to the patient's need, taking into account his/her medical history, social factors, ease of deliverance and patient expectations.

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