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Orthodontic–orthognathic surgical treatment of a subject with Williams–Beuren syndrome—a follow-up from 8 to 25 years of age

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SUMMARY This article presents a survey of characteristic features of Williams–Beuren syndrome (WBS) as reported in the literature and the interdisciplinary treatment of a subject with WBS with special regard to morphological and functional disorders.

Typical features of a patient with WBS and his dental development in a follow-up from 8 to 25 years of age are shown. Early orthodontic treatment approaches, later combined orthodontic–orthognathic surgical procedures, and the status 5 years after surgery are presented.

Introduction

The syndrome was first described by Williams *et al.* (1961). The subjects display a distinctive range of symptoms: supravalvular aortic stenosis, mental retardation, and dysmorphic facial features. Beuren *et al.* (1962) and Beuren (1964) independently described the syndrome noting also typical dental anomalies. The syndrome represents a developmental disorder caused by a hemizygous deletion of the elastin gene on chromosome 7q11.23 (Dutly and Schinzel, 1996; Donnai and Karmiloff-Smith, 2000). Williams–Beuren syndrome (WBS) occurs in approximately 1:20 000 live births (Morris *et al.*, 1988; Tarjan *et al.*, 2005) and is equally prevalent in both genders (Morris *et al.*, 1988).

Apart from diseases of specific tissue system (Table 1), WBS is characterized by typical dysmorphic facial features such as full prominent cheeks, wide mouth, long philtrum, and thick lips. These facial features are summarized in the term 'elfin face'. Furthermore, cephalometric measurements often show an anterior inclination of the maxilla, a high mandibular plane angle, and a deficient bony chin (Axelsson, 2005). These facial characteristics complicate treatment of the malocclusion, which would, in most cases, require an interdisciplinary approach.

Due to these facial dysmorphologies, early determination of treatment objectives and the timing of interdisciplinary strategies are important for adequate management.

Dental problems connected with WBS have received scant attention in the literature and no reports have been published with regard to orthodontic or orthognathic treatment.

The following presentation describes the characteristic morphological and functional disorders of the dentofacial complex in a patient with WBS leading to orthodontic–orthognathic treatment.

Subject and methods

Subject

An 8-year-old boy was referred for orthodontic consultation by his general dentist due to functional problems in connection with an anterior open bite (AOB).

Diagnostics

Anamnesis. The pregnancy, delivery, and perinatal experience of the mother had been uneventful. No medication with a known embryotoxic or foetotoxic potential had been taken during pregnancy. The family history did not reveal any further occurrence of WBS. The paediatrician suspected WBS at 8 years of age. Genetic testing by fluorescence *in situ* hybridization analysis was performed at 15 years of age and corroborated the clinical diagnosis based upon multiple typical signs of WBS (Table 1).

General clinical findings. The patient displayed unilateral renal aplasia. A pulmonary stenosis was diagnosed. Bilateral iris stellata (Figure 1), mild strabismus, and the typical hoarse voice were present. Furthermore, he was characterized by an overly friendly personality, being intellectually compromised.

Local clinical findings. The patient displayed the characteristic elfin face appearance including full prominent cheeks, wide mouth, long philtrum, small nose, and posteriorly rotated ears. Functional incompetent lips and impaired nasal breathing were diagnosed. Macroglossia with signs of lingua plicata (Figure 2) was combined with a severe tongue thrust. Oral hygiene was poor and in need of improvement; mild gingivitis and a high frequency of caries were prevalent.

Diagnostic records. Diagnostic records were taken at the following developmental stages:

T1: before introductory treatment at 8 years of age (Figures 3a, 4a, and 5a),

Table 1 Typical features of Williams–Beuren syndrome as reported in the literature and observed in the patient presented.

Manifestations	Literature	Patient presented
DNA aberration	Dutly and Schinzel (1996) and Donnai and Karmiloff-Smith (2000)	+
Unilateral renal hypoplasia	Ounap *et al.* (1998)	(Unilateral renal aplasia) +
Supravalvular aortic stenosis/cardiovascular anomalies	Williams *et al.* (1961); Morris *et al.* (1988); De Montgolfier-Aubron *et al.* (2000) Donnai and Karmiloff-Smith (2000); Axelsson *et al.* (2004); Axelsson (2005); and Tarjan *et al.* (2005)	(Pulmonal stenosis) +
Infantile hypercalcaemia	Williams *et al.* (1961); De Montgolfier-Aubron *et al.* (2000); Donnai and Karmiloff-Smith (2000); Tarjan *et al.* (2003); and Axelsson *et al.* (2004)	−
Hernia	Vernant *et al.* (1980)	(Inguinal hernia) +
Distinctive/overly friendly personality	Williams *et al.* (1961); Morris *et al.* (1988); De Montgolfier-Aubron *et al.* (2000); Donnai and Karmiloff-Smith (2000); and Axelsson *et al.* (2004)	+
Intellectual disability/ mental retardation	Williams *et al.* (1961); De Montgolfier-Aubron *et al.* (2000); Tarjan *et al.* (2003); Axelsson *et al.* (2004); Vicari (2004); and Tarjan *et al.* (2005)	+
Growth retardation/developmental delay	Morris *et al.* (1988); Donnai and Karmiloff-Smith (2000); Tarjan *et al.* (2003); Axelsson *et al.* (2004); and Axelsson (2005)	+
Short stature	Axelsson *et al.* (2004)	(1.65 m) +
Morphological aberration of the sella turcica	Axelsson *et al.* (2004)	−
Size aberration of the neurocranium	Axelsson (2005) and Axelsson *et al.* (2005)	(Scaphocephaly) +
Elfin face	Alberth *et al.* (1996); De Montgolfier-Aubron *et al.* (2000); Tarjan *et al.* (2003, 2005); and Axelsson (2005)	+
Full prominent cheeks	Metcalfe (1999) and Axelsson (2005)	+
Wide mouth	Metcalfe (1999) and Axelsson (2005)	+
Long philtrum	Axelsson (2005)	+
Thick lips	Tarjan *et al.* (2003)	+
Ocular findings (iris stellata, strabismus)	Holmstrom *et al.* (1990); De Ancos and Klainguti (1996); Winter *et al.* (1996); and Metcalfe (1999)	+
Small nose with depressed nasal bridge	Axelsson (2005)	+
Heavy orbital ridges	Axelsson (2005)	+
Medial eyebrow flare	Metcalfe (1999) and Axelsson (2005)	−
Hoarse voice	Metcalfe (1999) and Axelsson (2005)	+
Hyperacusis	Johnson *et al.* (2001)	−
Tongue thrusting	Hertzberg *et al.* (1994)	+
Anterior inclination of the maxilla	Axelsson (2005)	+
High mandibular plane angle	Axelsson (2005)	+
Deficient bony chin	Axelsson (2005)	+
Primary tooth resorption anomaly	Tarjan *et al.* (2003)	−
Aberrations in size, shape, and number of teeth	Beuren (1972); Hertzberg *et al.* (1994); Tarjan *et al.* (2003); Axelsson *et al.* (2004); and Axelsson (2005)	(Microdontia) +
Enamel hypoplasia	Hertzberg *et al.* (1994) and Alberth *et al.* (1996)	−

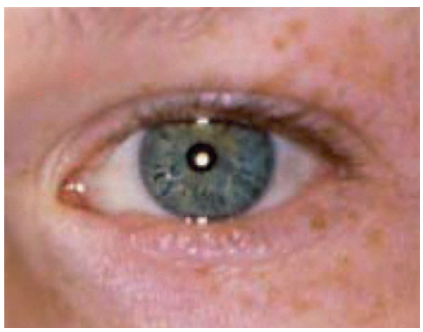

Figure 1 Iris stellata.

T2: before surgical treatment at 19 years of age (Figures 3b, 4b, and 5b), and
T3: 5 years after surgery at 25 years of age (Figures 3c, 4c, and 5c)

Initial diagnostic records (T1) included dental casts, a panoramic radiograph (Figure 5a), a lateral cephalometric headplate (Figure 4a), and intra- and extraoral photographs (Figures 3a and 5a).

Radiographic and model analysis (T1). The panoramic radiograph demonstrated a mixed dentition with multiple restorations and carious lesions. All permanent teeth were present. The stage of eruption between the permanent and primary teeth was normal according to age. Model analysis revealed an increased transverse width of the mandibular and maxillary arches, aberrations in shape and size of the teeth (microdontia), and, as a consequence, dental spacing. An Angle Class I molar relationship and an anterior crossbite combined with an AOB were diagnosed (overjet = −6 mm, overbite = −2 mm).

Cephalometric analysis (T1). Cephalometric analysis (Hasund, 1977) revealed a disharmonious orthognathic facial type with a neutral sagittal and an open vertical basal relationship (Table 2).

The vertical dimension displayed a distinct disproportion of the upper to lower anterior face height. A syndrome-related short upper anterior face height caused this

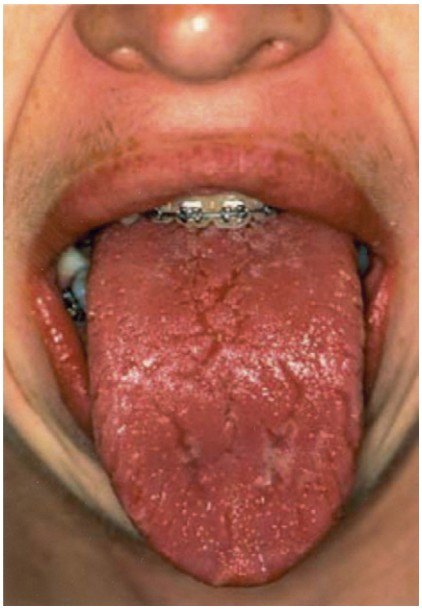

Figure 2 Macroglossia, lingua plicata.

disproportion and consequently the patient did not reveal a 'long face' appearance.

In addition, the vertical basal configuration indicated an anterior inclination of the maxilla. An unfavourable large gonial angle was responsible for the pronounced posterior inclination of the mandibular line and for the large interbasal angle. Furthermore, the mandible was characterized by a severely deficient bony chin.

The negative overjet was caused by slightly retruded upper incisors and severely protruded lower incisors (Figure 4a). The soft tissue analysis indicated that the lips exceeded the line of harmony (Holdaway angle) and thus caused a disharmonious extraoral appearance.

Growth prognosis. Growth prognosis predicted an indifferent translation and a posterior rotation of the mandible.

Therapy

Introductory treatment. Introductory functional treatment was started at 9 years of age. A modified Bionator was chosen in order to influence the dysfunction connected with the malocclusion. However, the patient found it difficult to use the appliance which did not remain in the mouth due to macroglossia. Only a minor reaction could be obtained and consequently the functional appliance therapy was discontinued.

Due to the functional problems in combination with the unfavourable vertical basal configuration, it was evident that conservative orthodontic treatment alone was insufficient. Thus, the main treatment was postponed for later combined orthodontic–orthognathic surgical treatment. In the meantime, continuous orthodontic observation was performed.

Puberty occurs early in WBS (Cherniske *et al.*, 1999; Partsch *et al.*, 1999); the patient reached his final body height (1.65 m) by 16 years of age. A hand – wrist radiograph taken at 17 years of age indicated the stage Ru.

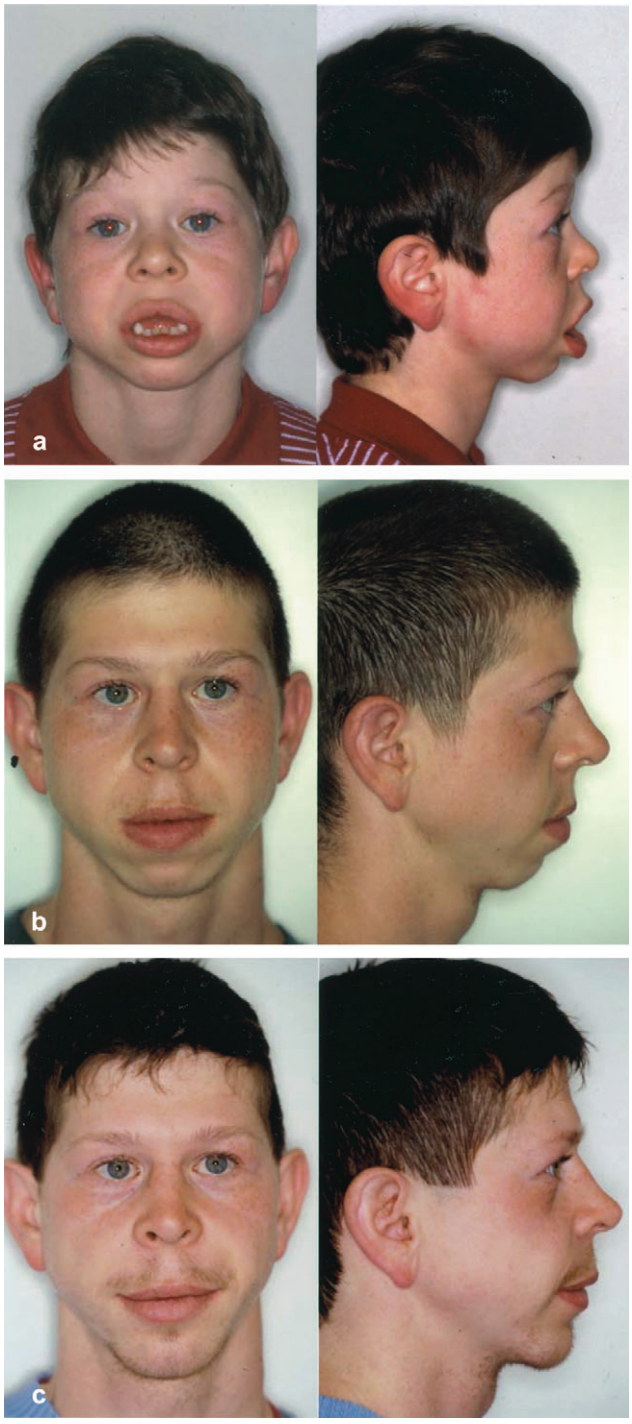

Figure 3 Extraoral view T1 (a), T2 (b), and T3 (c).

Main treatment. Diagnostic findings (T2) displayed a full permanent dentition with third molars erupted. Dental spacing, an Angle Class III molar and canine relationship, an anterior crossbite (overjet = −6 mm), and an AOB (overbite = −2 mm) were prevalent. The cephalometric changes (Table 2) were congruent with growth prognosis.

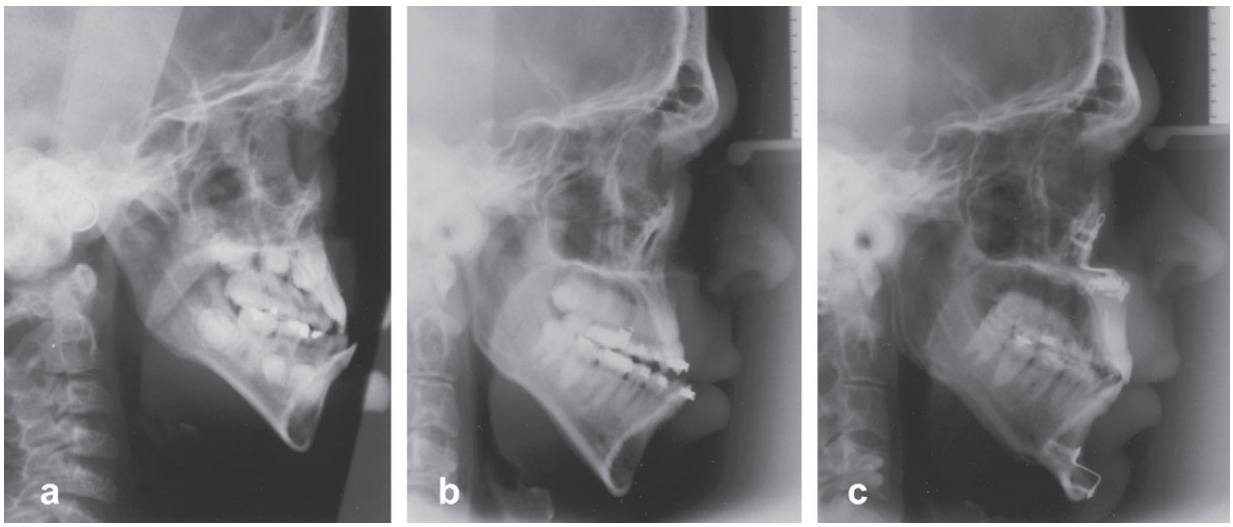

Figure 4 Lateral cephalometric radiographs T1 (a), T2 (b), and T3 (c).

Pre-surgical orthodontic treatment. Pre-surgical orthodontic treatment was carried out with a straightwire edgewise technique using a 0.018-inch high-torque system.

Dentoalveolar decompensation, space closure, and harmonizing the upper and lower arches were the main treatment objectives. No efforts were undertaken to close the AOB orthodontically in order to allow surgical rotation of the basal segments. Standard 0.016 × 0.022-inch steel arches were used for stabilization during and after surgery.

Orthognathic surgery. Orthognathic surgery was undertaken at 20 years of age. The maxilla was advanced at a Le Fort I level and divided into three pieces. The posterior parts of the maxilla were cranially positioned followed by a counterclockwise autorotation of the mandible in order to close the AOB. The maxilla was fixed with two angled Le Fort I miniplates applied laterally to the piriform aperture, one on each side. Simultaneously, a genioplasty was performed with a 10 mm advancement of the caudal chin segment. The chin was fixed with an angled miniplate. No intermaxillary fixation was used. One week after surgery, intermaxillary adaptation was supported by applying soft elastics according to the concept of semi-rigid bone fixation (Paulus, 1991).

Two months post-surgery the occlusion was Angle Class I with a well-defined overbite and overjet.

Post-surgical orthodontic treatment and retention. The healing took an uneventful course. Functional limitations or nerve disturbances did not occur. The miniplates remained *in situ*.

Orthodontic finishing after surgery was performed within 5 months by fixed appliances followed by removable retention plates for 1 year 6 months.

Long-term observation. At T3 there were no signs of relapse (Figures 4c and 5c). Molar and canine relationship was Angle Class I, the overbite and overjet were well defined, there was moderate spacing (due to microdontia), no disorders of the temporomandibular joint or muscular function, and no tongue thrust. An increasing tendency to caries development was observed. The patient was in good general health with regard to the syndrome.

Discussion

Systemic pathological features in WBS patients pose a challenge to orthodontic treatment. The syndromal, skeletal, and dental malformations and the additional tongue dysfunction require individualized, complex treatment planning.

Myofunctional therapy was not undertaken since it does not influence the aetiology for tongue thrust: the size of the tongue. Furthermore, retarded mental development limits excessive demands.

Macroglossia and tongue thrusting counteract, closing the AOB by conservative orthodontic treatment. An additional argument against conservative treatment might be the open basal configuration, mainly caused by an unfavourable mandibular structure. The patient displayed a high gonial angle, leading to an excessive posterior inclination of the mandible.

Interdisciplinary orthodontic–orthognathic treatment planning is required.

The following surgical interventions were taken into consideration: a partial glossectomy to reduce size of the tongue (Wolford and Cottrell, 1996), raising the mandible, and/or an advancement of the maxilla.

Partial glossectomy was rejected since potential risks and complications might occur including decreased movement of the tongue, residual speech, masticatory problems and anaesthesia of the tip of the tongue (Egyedi and Obwegeser, 1964).

With regard to long-term stability, the space for the tongue had to be enlarged. Consequently, the therapy of

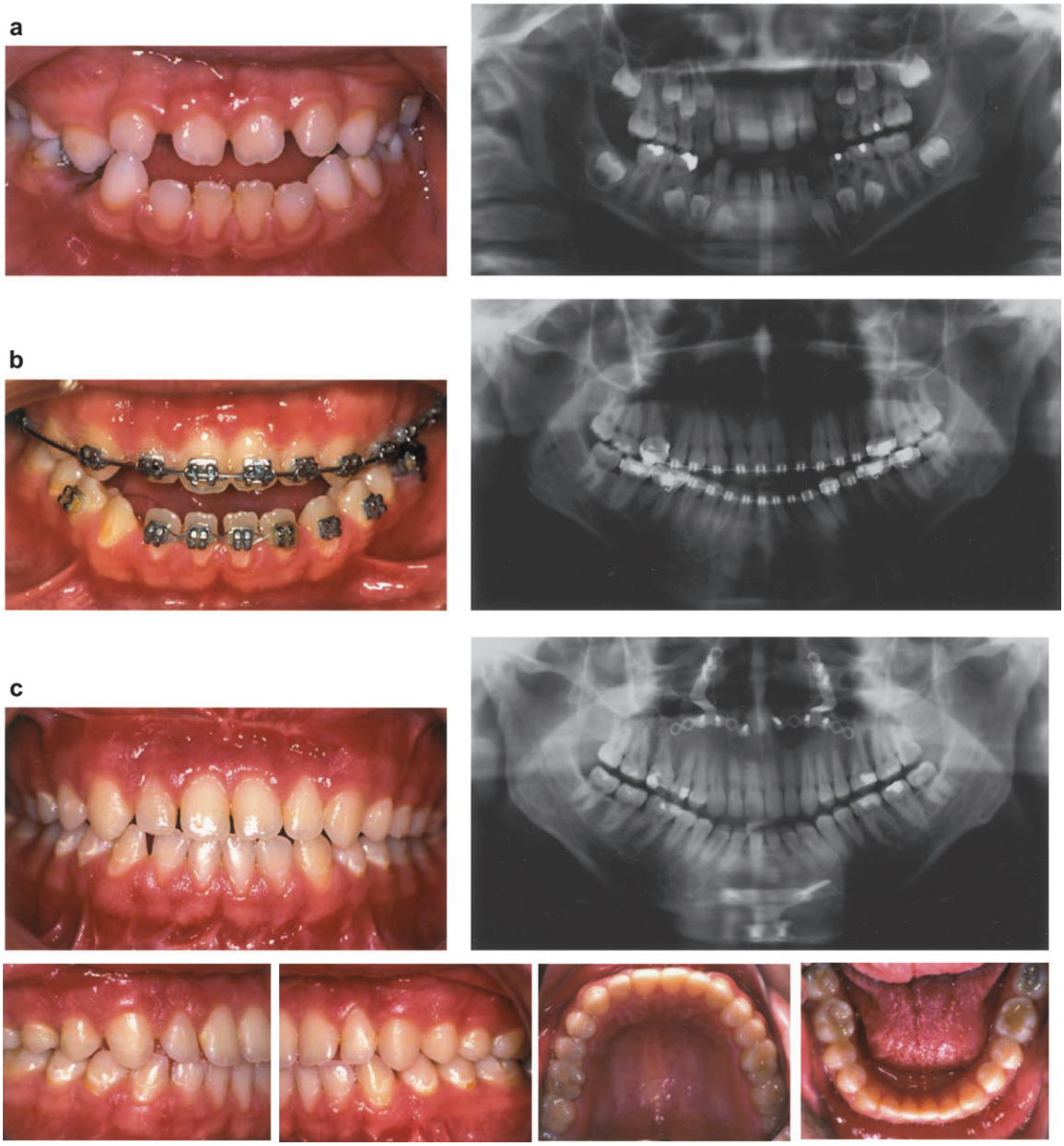

Figure 5 Intraoral views and panoramic radiographs T1 (a), T2 (b), and T3 (c).

choice comprised advancement and a posterior rotation of the maxilla and additional genioplasty.

The maxillary advancement and posterior rotation resulted in a reduction of the interbasal angle and led to maxillary prognathism, resulting in a distal sagittal basal relationship at T3 (Table 2). This morphological disharmony was intended to adapt the morphological structures to the functional demands: reducing tongue thrusting by enlarging the space for the tongue.

The genioplasty camouflaged the created sagittal discrepancy and harmonized the profile and facial aesthetics (Figure 6).

Despite an open vertical basal configuration, the chin was moved into an even more ventro-caudal position in order to harmonize the total anterior face height in relation to the transverse facial width characterizing the elfin face appearance.

The post-surgical appearance led to an increase in the patient's self-confidence and improved social acceptance.

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Table 2 Cephalometric measurements.

Measurements	Mean	T1	T2	T3
SNA (°)	82	82.5	81	87.5
SNB (°)	80	80.5	79.5	80.5
ANB (°)	2	2	1.5	7
SNPg (°)	81	78.5	77.5	82
NSBa (°)	130	131.5	131.5	132
Gn-tgo-Ar (°)	126	153	153	153
H angle (°)	8	19.5	21	18
ML-NSL (°)	32	47	49	45.5
NL-NSL (°)	8.5	6.5	9.5	12.5
ML-NL (°)	23.5	40.5	39.5	33
N-Sp′ (mm)		45	51	51
Sp′-Gn (mm)		66	78.5	80
Index (%)	79	68.2	65	63.8
1-1 (°)	131	117	125.5	125
1-NA (°)	22	18	17.5	10
1-NB (°)	25	43	36	37
1-NA (mm)	4	15	3.5	−1
1-NB (mm)	4	9	11	9
Pg-NB (mm)	2	−4	−4	3.5

T1: before treatment at 8 years of age; T2: before surgical treatment at 19 years of age; T3: 5 years after surgery at 25 years of age.

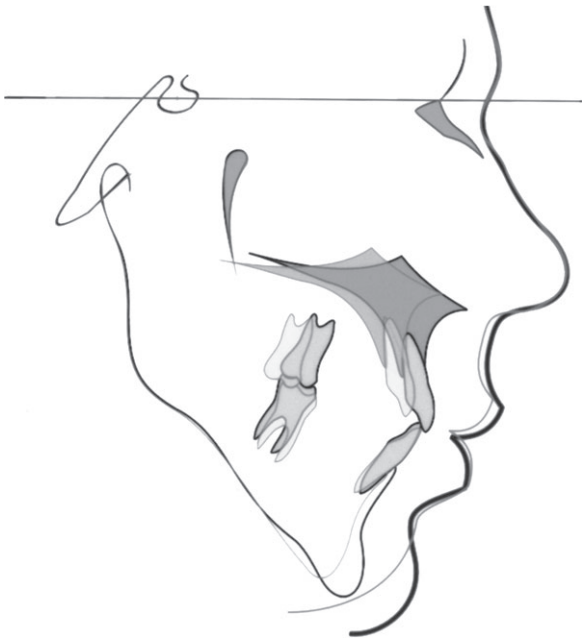

Figure 6 Superimposed cephalometric tracings T2 (grey line) and T3 (black line).

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