Case Report

Orthodontic Treatment Protocol of Ehlers-Danlos Syndrome Type VI

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Abstract: Ehlers-Danlos syndrome (EDS) type VI is an autosomal recessive disorder of the connective tissue characterized by joint hypermobility, muscle hypotonia, scoliosis, and ocular fragility. In this case report, an EDS type VI patient with a skeletal and dental Class III malocclusion is presented and the clinical approach to his orthodontic problem is emphasized. A 17-year-old male patient presenting some major and minor symptoms of the syndrome was referred to our orthodontic department for diagnosis and treatment. The typical clinical signs confirmed the diagnosis of EDS type VI. He was a skeletal and dental Class III malocclusion patient (both mandibular protrusion and maxillary retrusion) with a noncontributory family history. He had severe crowding in the lower and upper dental arches with retruded incisors. His first treatment plan included orthognathic surgery, but because of the risks of bleeding and poor healing, we elected to treat the patient without surgery. (*Angle Orthod* 2006;76:177–183.)

Key Words: Ehlers-Danlos syndrome; Connective tissue disorder; Missing frenula; Class III malocclusion

INTRODUCTION

Ehlers-Danlos syndrome (EDS) is a group of generalized disorders characterized by abnormalities of the connective tissue leading to fragility of the skin and blood vessels, hyperextensibility of the skin, and joint hypermobility.[1–3] The prevalence is about 1 in 5000 births.[4] The classification of the syndrome is according to the major and minor diagnostic criteria. On the basis of these clinical symptoms, there are at least 10 types of the syndrome. However, about half of the patients with EDS do not fit into 1 of the 10 types.[5] In 1997, Beighton et al[6] revised the classification because of recent developments in the elucidation of the biochemical and molecular bases of EDS, together with in-

creasing clinical experience. Some forms are uncommon (EDS types IV, VI, VII, and VIII),[7] whereas others have been found in only a few families (EDS types IX and X).[4]

EDS type VI is an autosomal recessive disorder.[6,8] The major diagnostic criteria are general joint hypermobility, muscle hypotonia, scoliosis, and ocular fragility. In addition to these symptoms, the minor diagnostic criteria are tissue fragility, atrophic scars, easy bruising, arterial rupture, tricuspid insufficiency, marfanoid habitus, microcornea, considerable radiological osteopenia, and family history.[3,6,8,9] In cases with a progressive kyphoscoliosis, thoracic cage deformity and hypotonia lead to a decrease in pulmonary function and favor recurrent episodes of pneumonia, relative coronary insufficiency, and early death.[10] The diagnosis is made by finding decreased amounts of hydroxylysine in skin and confirmed by low levels of lysyl hydroxylase measured in cultured dermal fibroblasts.[4,8]

Absence of the inferior labial and lingual frenula was found to be an oral manifestation associated with classical and hypermobility types of the syndrome.[11] Other dental symptoms include fragile oral mucosa, early onset of periodontal defects, unusual dental crown anatomy, dental fractures, stunted roots or dilacerations, aberrant dentinal tubules, pulpal vascular lesions and denticles, hypermobile temporomandibular joint (TMJ), and ready movement of teeth in response to orthodontic treatment.[12]

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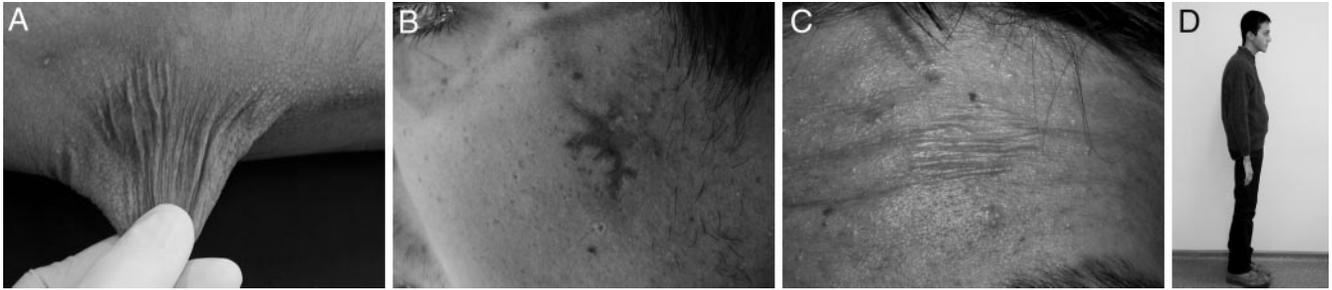(e-mail: dnalbantgil@yeditepe.edu.tr)

FIGURE 1. (a–d) Major and minor diagnostic criteria of the syndrome.

Some patients with EDS type VI respond to ascorbic acid (one to four g/day), with some symptomatic improvement and increased excretion of hydroxylysine in the urine.[4] There is no metabolic treatment for other forms of EDS, and management is largely symptomatic.[4]

In this case report, an EDS type VI patient with a skeletal and dental Class III malocclusion is presented and the clinical approach to his orthodontic problem is emphasized.

CASE REPORT

Diagnosis and etiology

A 17-year-old male patient with skeletal and dental Class III malocclusion was referred to our Department of Orthodontics for diagnosis and treatment. According to his patient history, he was born at week 40 of the pregnancy. His birth weight was 2250 g and length was 52 cm. His mother, a nurse, noticed his muscle weakness at birth. He was a weak baby, and his motor development was delayed (walking at 3 years of age). On the other hand, he started talking when he was 2 years old, and his intellectual development was normal. At the age of 4 years, further physical symptoms evolved, such as luxation of his shoulder and development of strabismus, which led him to wear glasses.

The patient came to our clinic with a diagnosis of EDS type VI and presented the clinical diagnostic features of the syndrome. His clinical dermatological examination revealed soft, velvety, and hyperextensible skin (Figure 1a). Easy bruising, atrophic, and ''cigarette-paper'' scars were present in the areas of trauma (Figure 1b,c). He had hypotrophic muscles, habitual luxation of right shoulder, and joint hypermobility, which tend to decrease after puberty. In addition to these signs and symptoms, he had a marfanoid habitus with pectus excavatum, hallux valgus, and long extremities (Figure 1d). As one of the major criteria of EDS type VI, he had scoliosis, and his radiograph showed osteopenia. His cardiological examination revealed moderate tricuspid insufficiency. Keratoconus, ocular fragility, narrow visual field, and microcornea

FIGURE 2. Oral manifestation; missing inferior labial and lingual frenula.

were his ophthalmologic diagnostic criteria. He had undescended testicles, which were surgically removed in a simultaneous event unrelated to his orthodontic treatment.

As an oral manifestation, his inferior labial and lingual frenula were missing (Figure 2). Other than this, he had crenulated incisors and high cusps and deep fissures on the crowns of the teeth. The patient's chief complaint was severe crowding in both dental arches. Clinically, he had a long face, a constricted maxilla with bilateral posterior crossbite, high smiling line, and a straight profile with concave paranasal areas (Figure 3a through c).

Cephalometric analysis revealed a skeletal Class III relationship (ANB − 3.7°, both mandibular protrusion and maxillary retrusion) with a noncontributory family history. He had a hypodivergent facial pattern, and a severe transverse deficiency (bilateral crossbite) was noted in the maxilla.

Model analyses showed a Class III malocclusion with a very narrow maxilla. His overjet and overbite were −0.8 and −0.3 mm, respectively. He had 8 mm of crowding in the mandibular arch, and his lower incisors were in a retroclined position. In the maxilla, he had an arch discrepancy of −12.5 mm, and his upper incisors were retruded. He had no Bolton discrepancy.
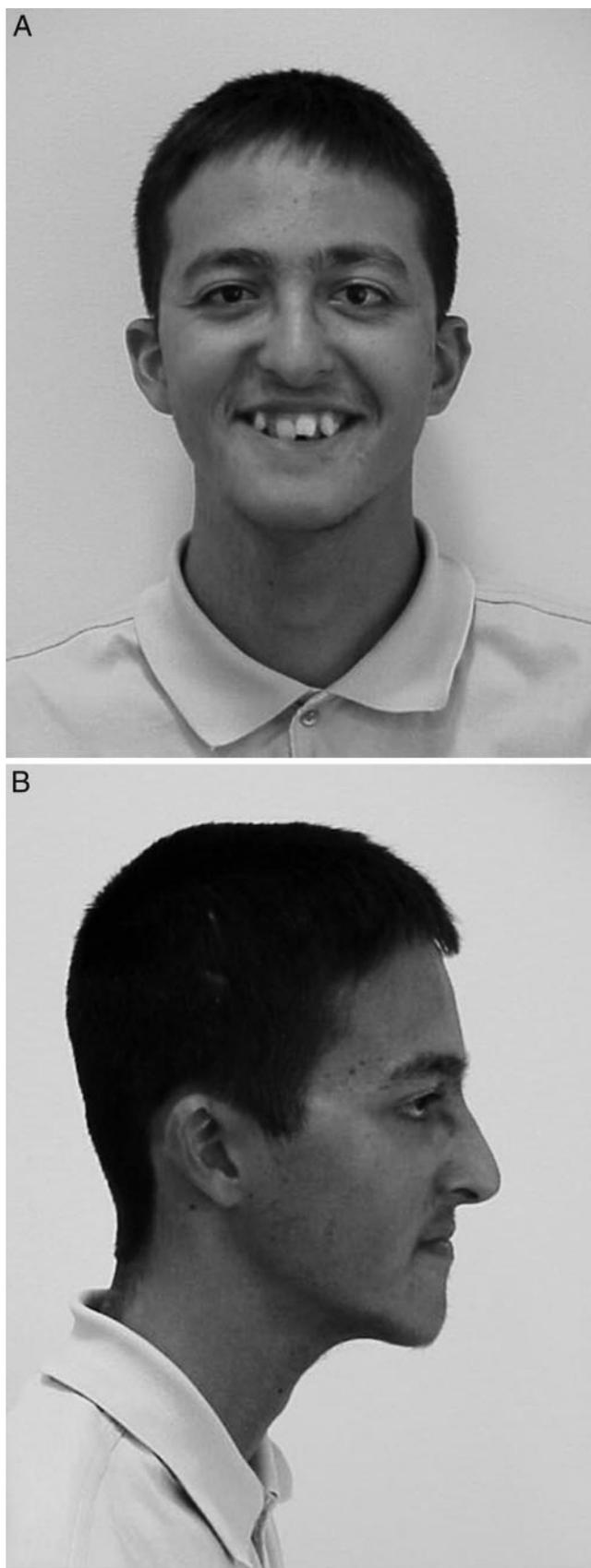
FIGURE 3. (a–b) Pretreatment extraoral photographs.

Because of transverse maxillary deficiency, bilateral posterior and anterior crossbites were exhibited. The lower dental midline was 1.5 mm to the right in relation to the facial midline (Figure 4a through e).

The panoramic radiograph revealed normal root morphology. All his wisdom teeth were present, and his right upper first molar had been extracted. He had a supernumerary tooth at the apex of the right upper central incisor (Figure 5). Because he had no history of TMJ hypermobility, closed and open tomography and arthromograms were considered unnecessary. Contrary to what might have been expected with EDS type VI, the patient presented good periodontal health and no recession or gingival bleeding.

Treatment alternatives and objectives

His initial treatment plan included orthognathic surgery, but because of the risks of bleeding and poor healing, we elected to treat the patient without surgery.

Because he had a severe transverse deficiency in the upper jaw, rapid palatal expansion had to be performed to obtain enough space and to develop a well-shaped archform. To correct the crowding along with the dental midline in the lower arch, the lower left central incisor was extracted after a model setup was made. The teeth located in the lower right anterior segment were believed to be shifted mesially, which would result in reshaping the lower right canine to become the lateral incisor and the first premolar to serve as the canine by selective grinding. Maxillary and mandibular incisors were to be proclined to an optimal overjet and overbite relationship.

Treatment progress

Because the patient had abnormalities of the connective tissue, which lead to poor healing, special care was given to his oral hygiene and periodontal status. He was observed at 3-month intervals by a periodontist to keep his periodontal status under control and stable throughout the treatment.

The expansion appliance was an acrylic splint type (bonded hyrax), which covered the upper posterior teeth. The patient activated the screw one-quarter turn every 12 hours. Within a week, the midpalatal suture was opened. After the expansion was completed, his impacted supernumerary tooth was surgically extracted. Standard Edgewise brackets (0.018 in) were placed on the anterior teeth and the first premolars by skipping the upper left lateral incisor. Beta III Titanium alloy (CNA) segmental archwire (0.016 $\times$ 0.022 in) with loops was used for leveling as the initial wire and was activated forward to eliminate the negative overjet at every 4-week recall appointment for 4 months (Figure 6a through d).
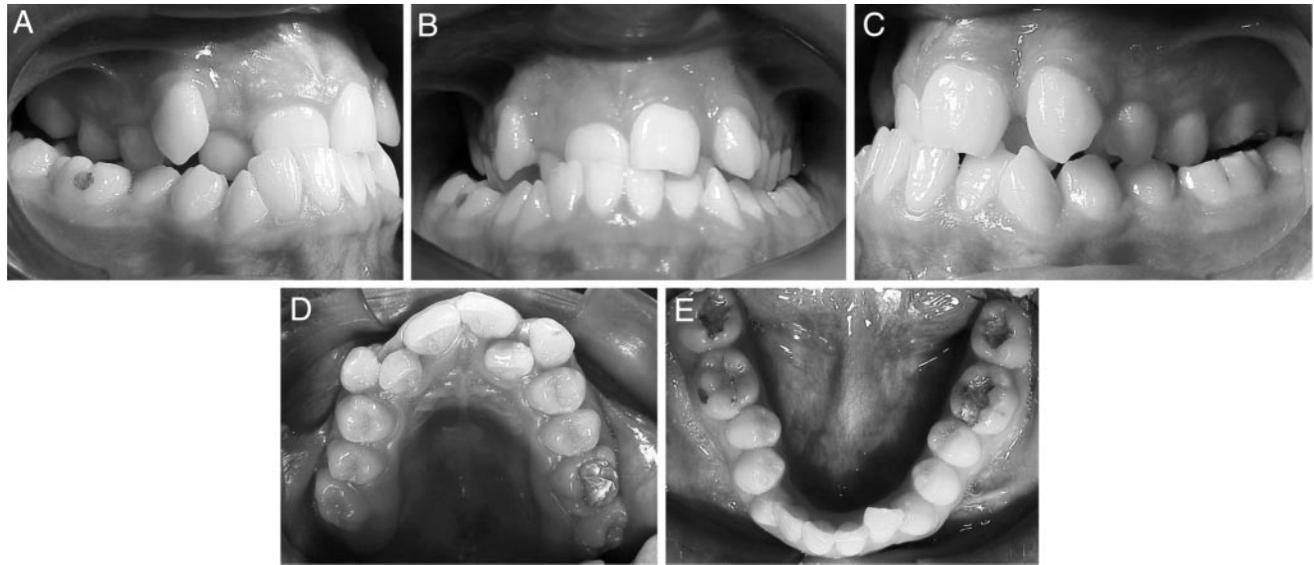
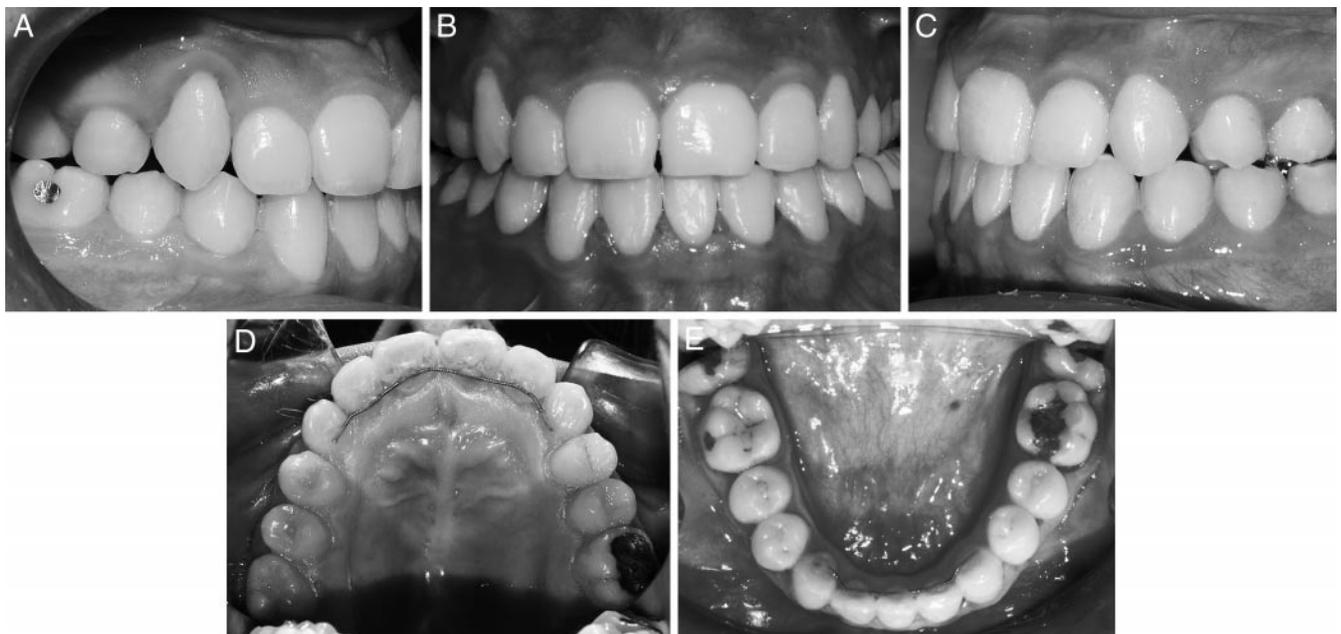**FIGURE 4.** (a–e) Pretreatment intraoral photographs.

FIGURE 5. (a–e) Posttreatment intraoral photographs.

To provide the necessary space to align the lower dental arch, the lower left central incisor was extracted after a laboratory study cast setup model was made. After the extraction, brackets were placed on the posterior teeth, including the canines, and sectional 0.014-in NiTi archwires were used for leveling. The extraction space closure was started one month later using light forces.

The hyrax appliance was removed 7 months after the expansion was completed, and a transpalatal arch was placed to prevent relapse. The posterior brackets were placed and a 0.016 × 0.016–in NiTi archwire with molar stops was used to protrude the upper incisors. Eleven months after the treatment had initiated, the patient was out of anterior crossbite by protrusion of the maxillary anterior teeth.

After the leveling stage, Class III elastics were used on 0.016 × 0.022–in CNA archwires to obtain an optimal occlusion. Toward the end of the treatment, the proximal sides of the crenulated upper incisors were ground to obtain better contact surfaces and more pleasing and esthetic incisor shapes. After the com-
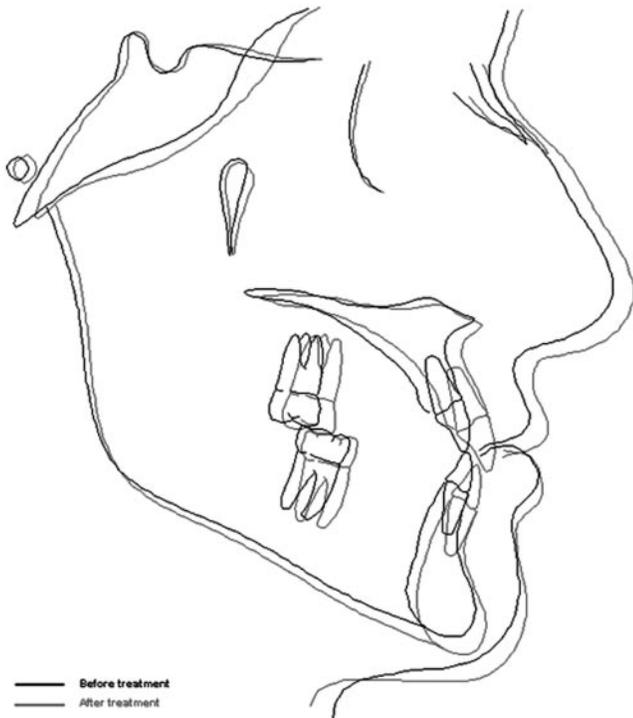
FIGURE 6. Posttreatment lateral cephalometric tracing superimposed on pretreatment tracing.

pletion of orthodontic intervention, which lasted 34 months, the modification of the displaced teeth was performed. The cusp and palatal surface of the lower right canine was ground and flattened. Similarly, slight grinding was performed to the lingual cusp of the first premolar to prevent occlusal interference.

Permanent retention was used with a lingually bonded multistranded wire over six teeth in the upper and five teeth in the lower jaw.

RESULTS

A satisfactory result was achieved in this patient with a compromise orthodontic treatment (Figure 5a through e). Clinical and radiographic records were obtained before, during, and after treatment. Evaluation of the posttreatment panoramic radiograph revealed no unusual root resorption.

The maxillary constriction was corrected with a bonded hyrax within seven months and enough space and well-shaped arch form were obtained in the maxillary arch. The surgical extraction of the impacted supernumerary tooth was performed after the expansion was completed to facilitate the surgeon's job. The arch deficiency in the mandible was corrected by extracting an incisor. Maxillary and mandibular incisors were proclined to an optimal overjet and overbite relationship. Although the lower left central incisor was extracted, the upper and lower dental midlines were co-
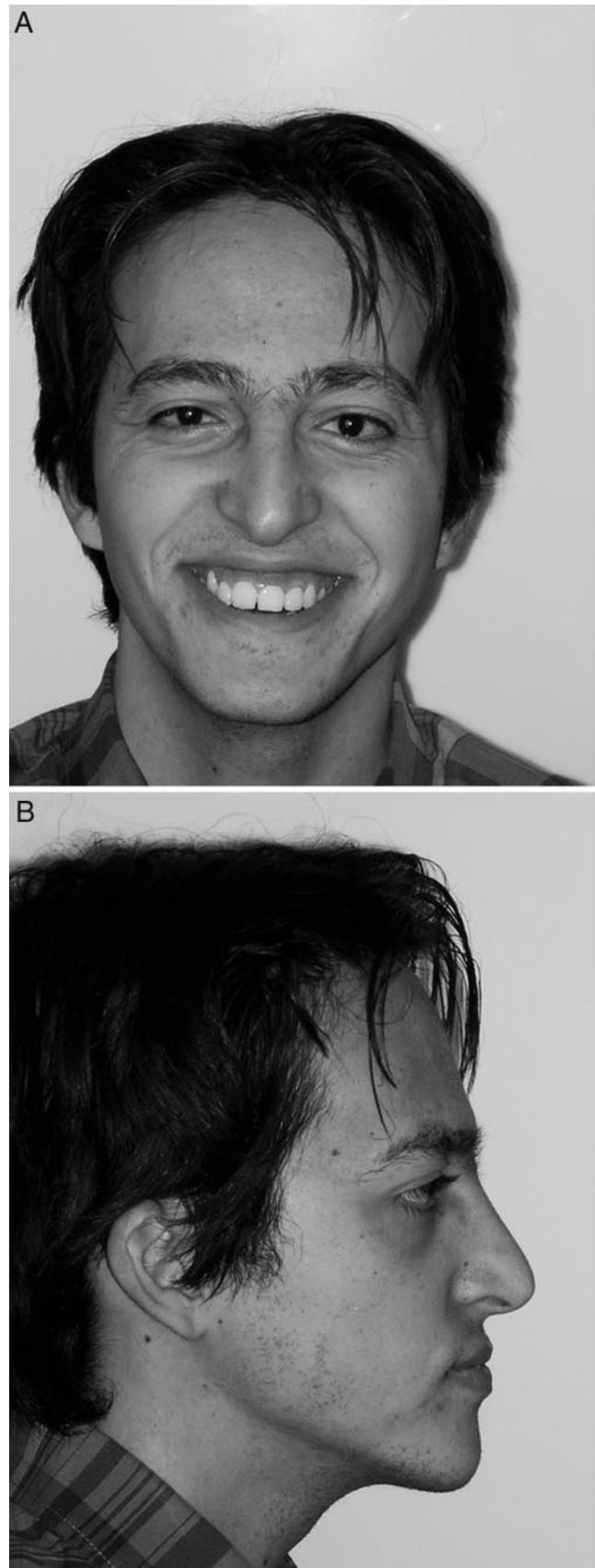

FIGURE 7. (a–b) Posttreatment extraoral photographs.

TABLE 1. Cephalometric Measurements of Pretreatment and Posttreatment Values

	Pretreatment (16 y nine mo)	Posttreatment (19 y seven mo)
Skeletal		
SNA (°)	86.3	87.4
SNB (°)	90.1	89.5
ANB (°)	−3.7	−2.1
MP-SN (°)	25.8	26.5
Sum of angles (%)	385.8	386.5
ANS-Me/Na-Me (%)	55.2	55.7
Jarabak ratio (%)	72.5	71.2
FMA (°)	21.9	22.2
Y-axis	56.7	56.2
Occ plane to SN (°)	7.4	10.4
SN-palatal plane (°)	4.9	5.3
Dental		
U1-SN (°)	111.4	110.8
IMPA (°)	76.6	78.5
Pog-NB (mm)	4.4	5.2
Interincisal angle (°)	146.3	144.2
Overjet (mm)	−0.8	2.8
Overbite (mm)	−0.3	1.6
Soft tissue		
H-angle (°)	6.6	9.0
Soft tissue convexity (°)	133.9	131.4
Nasolabial angle (°)	108.2	99.8

incident with each other and the facial midline at the end of the treatment.

Cephalometric measurements of pretreatment and posttreatment values are shown in Table 1. The sagittal relationship between the upper and the lower jaws was improved by a decrease in the ANB angle. Slight increase was detected in the SNGOGn angle (0.7°). The soft tissue profile improved significantly by the reflection of the changes that took place in the dentoskeletal structures (Figure 6). Profile harmony was established by supporting the soft tissue on paranasal areas by advancement of the underlying dentoalveolar structures (Figure 7a–b).

DISCUSSION

The clinical diagnostic features of the syndrome were observed in the patient of our study when he first came to the clinic, so an overall physical and dental consultation was needed before orthodontic treatment. The evaluation of the model and cephalometric analysis revealed an initial treatment plan, which included orthognathic surgery. However, because of the risks of bleeding and poor healing, and even the possibility of the presence of the late complications, which may include vascular rupture because of his disease,[4] a surgical treatment was not considered.

His compromise treatment plan included expansion of the maxillary arch, surgical extraction of the im-

pacted supernumerary tooth, protrusion of the upper anterior incisors, and extraction of the lower incisor to obtain enough space for the mandibular arch. As one of the cardinal features of the symptom, the patient had habitual luxation of his right shoulder and joint hypermobility. Although he did not have a history concerning the TMJ, the risk of subluxation of this joint remained throughout the treatment because the laxity of the supporting ligaments of the joint also permit subluxation and dislocation during opening.[12] Therefore, special care was taken that the patient not open his mouth wide during appointments, and a clinical examination of the TMJ was performed every 6 months.

Because of another major symptom of the syndrome, the patient had a tendency to slow healing, fragile oral mucosa, and early onset of periodontal defects. Thus, his oral hygiene and periodontal status were kept under control by periodontal checkups, which were conducted with 3-month recall visits by a periodontist, during his orthodontic treatment.

The patient had an excessive transverse maxillary deficiency with bilateral posterior and anterior crossbites, which necessitated palatal expansion. A large amount of maxillary expansion was accomplished with a bonded hyrax. Precaution was taken during fabrication for the appliance to have smooth edges so as not to abrade the tongue and buccal mucosa because the patient had problems with tissue repair and fragile oral mucosa.

Clinical and radiographic evaluation confirmed that the midpalatal suture was opened within a week. The result was obtained quickly because in EDS patients, tooth movement is expected to be more rapid for a constant appliance activation.[12] However, during the tooth movement stage, the mobility of the teeth may be greater than normal. The mobility may be caused by the stressed periodontal fibers on the tension side being stretched, torn, or rendered slow in repair.[12] Therefore, light forces were used throughout the treatment to avoid any untoward effects.

Because the tooth movement was accomplished quickly and easily due to the underlying connective tissue disorder, and the amount of the maxillary expansion achieved is large, permanent retention was preferred at the end of the treatment.

CONCLUSIONS

- Satisfactory results can be achieved by knowing the general and dental manifestations of the syndrome and taking appropriate precautions during orthodontic treatment.

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