Case Report

Orthodontic Treatment in a Patient with Cleidocranial Dysostosis

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ABSTRACT

Cleidocranial dysostosis is a rare congenital skeletal disorder, associated with clavicular hypoplasia or aplasia, delayed closure of cranial fontanels, brachycephalic skull, delayed exfoliation of primary dentition, eruption of permanent teeth, and multiple supernumerary and morphologic abnormalities of the maxilla and mandible. The disorder is caused by mutation in the CBFA1 gene, on the short arm of chromosome 6p21. The prevalence of cleidocranial dysostosis is estimated one per million, without sex or ethnic group predilection. The purpose of this paper is to describe the orthodontic treatment in a patient with cleidocranial dysostosis. Therapy may include removal of supernumerary teeth, surgical exposure of impacted teeth, and orthodontic treatment. (*Angle Orthod.* 2009;79:178–185.)

KEY WORDS: Cleidocranial dysostosis; Hypoplastic or aplastic clavicles; Supernumerary teeth

INTRODUCTION

Cleidocranial dysostosis (CCD) is a rare congenital disorder of bone with an autosomal dominant hereditary mode of inheritance. This condition is characterized by clavicular aplasia or deficient formation of the clavicles, delayed and imperfect ossification of the cranium, moderately short stature, and a variety of other skeletal abnormalities. The oral manifestations are a delayed exfoliation of primary teeth, delayed or failing eruption of the permanent dentition, and multiple supernumerary teeth.

The first case of clavicular defects was reported by Martin in 1765.[1] Another case with both clavicles and the skull affected was reported in 1871 by Scheuthauer.[2] In 1897 Marie and Sainton[3] coined the descriptive title cleidocranial dysostosis. The condition was originally thought to involve bones of intramembranous origin only, namely the bones of the skull, clavicles and flat bones, hence the name cleidocranial. Hesse[4] was first to describe in detail the defects of dentition and jaws associated with cleidocranial dysostosis. Yamamoto et al reported an individual with 63 supernumerary teeth.[5]

The gene for cleidocranial dysostosis has been mapped on the short arm of chromosome 6p21, core binding factor α-1 (CBFA1).[6–8] This disorder can be caused by mutation in the transcription factor CBFA1 (RUNX2). The CBFA1 gene controls differentiation of precursor cells into osteoblasts and is thus essential for membranous as well as endochondral bone formation, which may be related to delayed ossification of the skull, teeth, pelvis, and clavicles.[9] Yoshida et al[10] used genotype-phenotype studies to show that skeletal growth and dental development could be related to the type of mutations in the RUNX2 gene.

Zheng et al[11] concluded that the patients with cleidocranial dysostosis have altered endochondral ossification due to altered RUNX2 regulation of hypertrophic chondrocyte-specific genes during chondrocyte maturation.

The prevalence of cleidocranial dysostosis is one per million with complete penetrance and variable ex-

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Table 1. Clinical Findings in Cleidocranial Dysostosis

Hypoplasia clavicles	Aplasia clavicles
Brachycephalic skull	Midface hypoplasia
Delayed closure of fontanels	Thickness of the calvaria of the skull
Short stature	Arms and legs small
Genua valga	Pes planus
Scoliosis	Osteoporosis
Asthma	Hearing loss
Delayed exfoliation of primary teeth	Delayed eruption of permanent teeth
Supernumerary teeth	Mandibular prognathism
Maxillary hypoplasia	Dentigerous cysts
High palate	Cleft palate
Class II malocclusion	Class III malocclusion
Alteration of dental hard tissue	

pressivity, but it is most likely under diagnosed because of the relative lack of medical complications in comparison with other skeletal dysplasias.[9,12] Early diagnosis of CCD is essential for introduction of appropriate treatment approach based on interdisciplinary cooperation between orthodontists as well as oral and maxillofacial surgeons.

It may be discovered at any age, but the cranial deficiencies may be noticed at birth. Both sexes are affected to an approximately equal extent. The defect often appears in several successive generations.

The most characteristic and pathognomonic feature of this disorder is hypoplasia or aplasia of the clavicles, which results in hypermobility of the shoulders allowing the patients to approximate the shoulders in the midline. Muscle attachments to the clavicles may also be dysplastic, leading to distortion of the neck. Defects of the cervical and lumbar vertebrae are included in the clinical findings. Absence of the pubic symphysis and hypoplasia of the pelvis is common in female individuals. Frequently, genua valga and pes planus are found in children younger than 5 years of age.[13]

Moderately short stature was observed. The arms and legs are abnormally short; furthermore, the defects are revealed in hands, feet, and nails.[14]

The skull and facial appearance may also be characteristic. Delayed ossification of the cranial sutures and fontanels occurs, and may remain open throughout life. The skull is usually large and broad, like a brachycephalic type.

The face appears small in relation to the cranium with hypoplastic maxillary, lachrymal, nasal, and zygomatic bones. The paranasal sinuses may be underdeveloped. The bones of the middle part of the face are less well developed than the cranial bones. Defects in the skull appear to be always symmetrical. The frontal, parietal, and occipital bones are prominent. The maxillary sinuses may be small or missing, and the maxilla is underdeveloped, causing a relative mandibular protrusion.

The palate may be abnormally high, and occasionally cleft palate has been reported. Ocular hypertelorism and mild exophthalmus are seen. These individuals have short stature. Postural defects and spinal curvature are common.

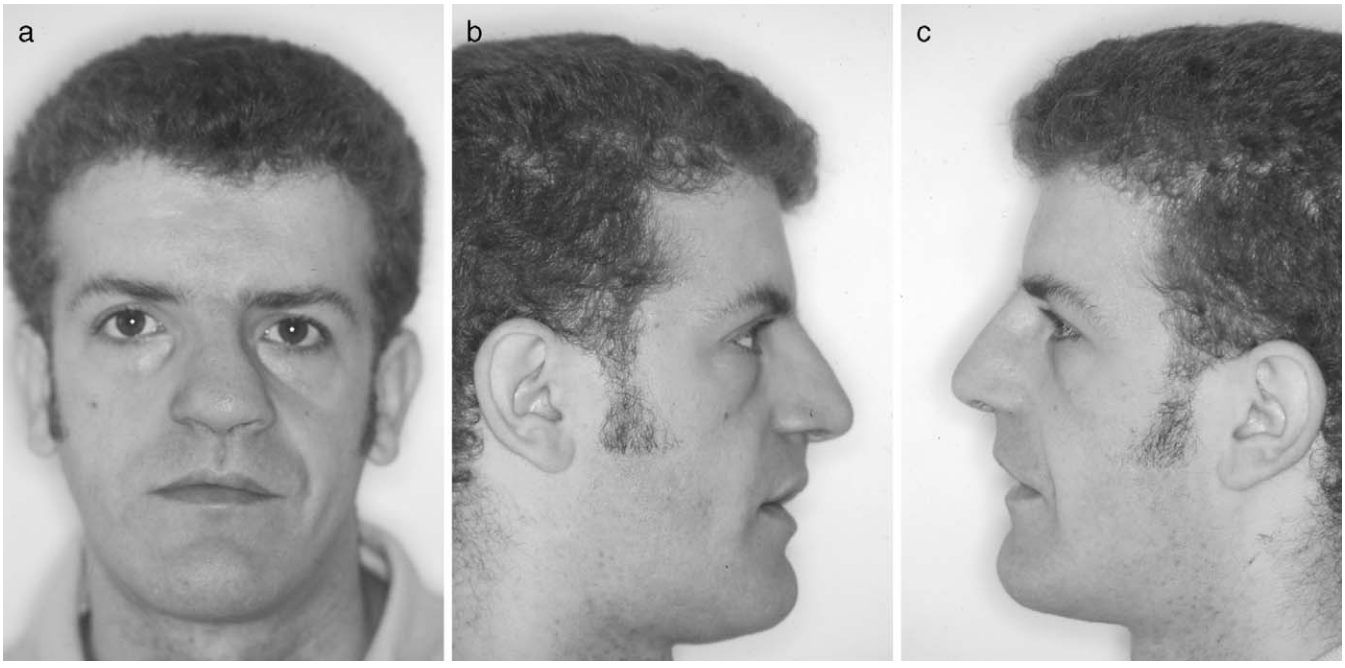

Figure 1. (a–c) Lateral and frontal view of the patient with cleidocranial dysostosis (CCD).

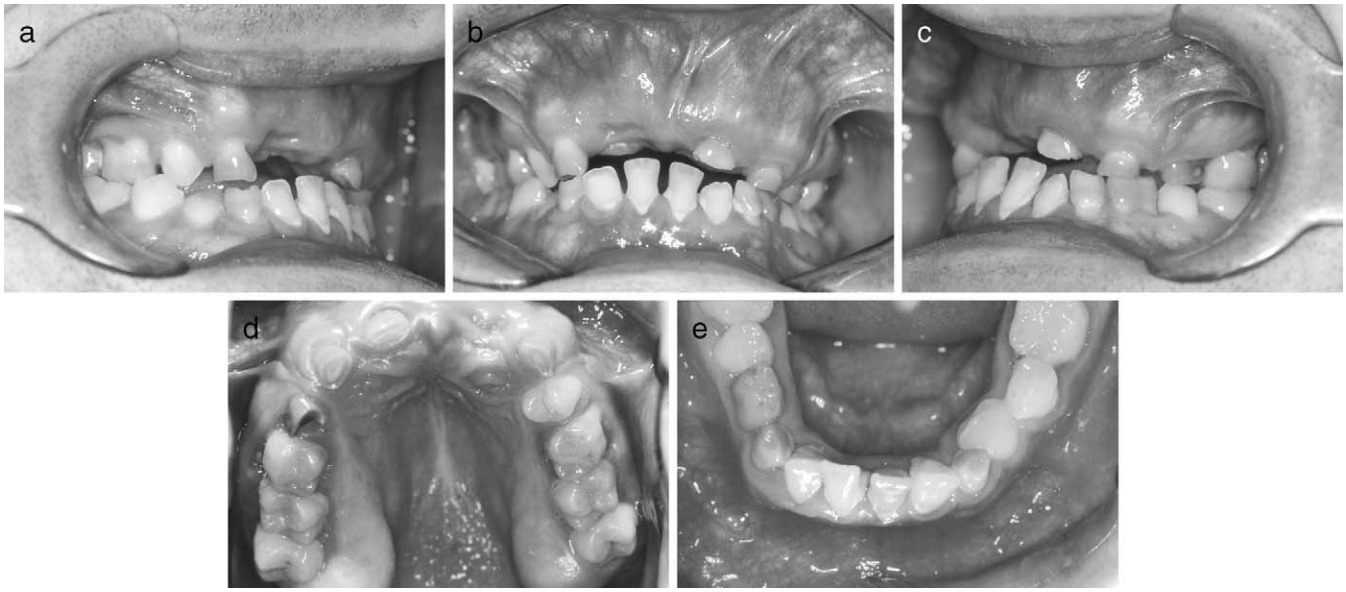

Figure 2. (a–e) Intraoral and occlusal view.

In the hands and the feet, various abnormalities have been found, the most constant and curious being the presence of epiphyses at both ends of the metacarpals and metatarsals, particularly of the second and fifth, and an abnormally long second metacarpal. The intermediate phalanges may be small. Ossification of the carpus may be delayed. Association with mental retardation has been shown, but most patients apparently possess normal intelligence. These individuals have no significant physical handicap.

The eruption of primary teeth is normal or sometimes delayed, but the exfoliation of primary teeth is always delayed and may be due to the failure of most permanent teeth to erupt. The presence of supernumerary teeth has been hypothesized to result from incomplete or delayed resorption of the dental lamina.[4,15] Dental manifestations include delayed eruption or failure of eruption of the primary dentition (Table 1). Delayed tooth development has been reported in association with malocclusion and supernumerary teeth. Dentigerous cysts may occur. Alterations of hard dental tissues and a high propensity for caries have been found.

The significant variability in clinical expression of this syndrome reflects a degree of phenotypic polymorphism.

MATERIALS AND METHODS

The aim of this paper is to describe the orthodontic treatment of a 28-year-old white male patient with CCD. His chief complaint was the failure of eruption of the upper permanent central incisor. He suffered

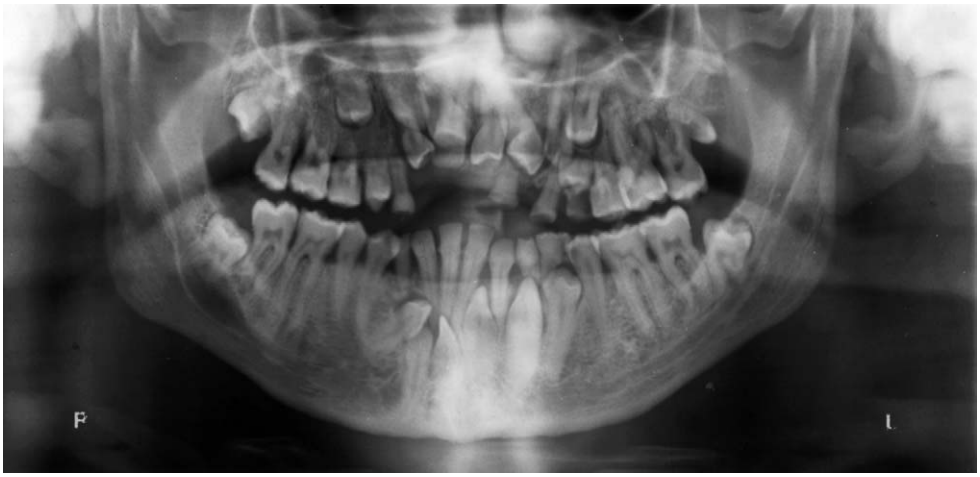

Figure 3. Panoramic x-ray demonstrating the dental condition.

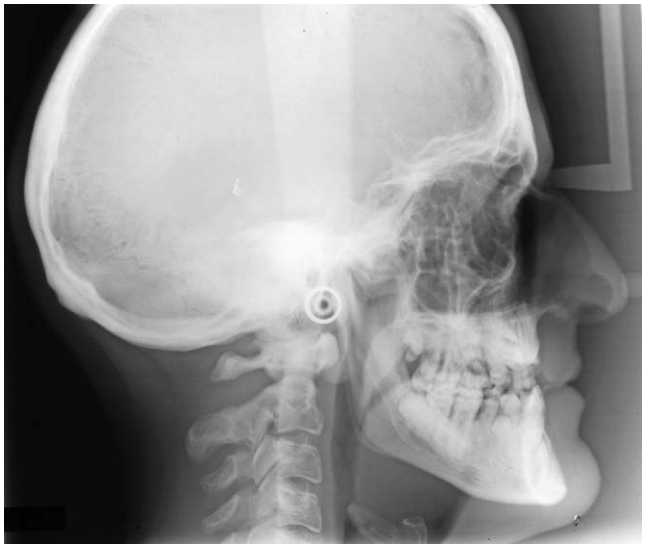

Figure 4. Lateral radiograph view.

from disabilities in eating, due to the distribution and eruption of anomalous teeth, dental esthetics, and facial appearance. A family history did not reveal similar signs or a hereditary disorder on either side of the family. There was no parental consanguinity.

Diagnosis was based on the bilateral hypoplasia of the clavicles, the presence of an enlarged cranium, frontal bossing, depressed suborbital region, defective nasal bones, failure of eruption, and multiple supernumerary teeth. This patient had a moderately short stature and a history of slow growth. Cognitive development was entirely within normal limits. The craniofacial findings included delayed closure of cranial fontanels and suture and brachycephalia.

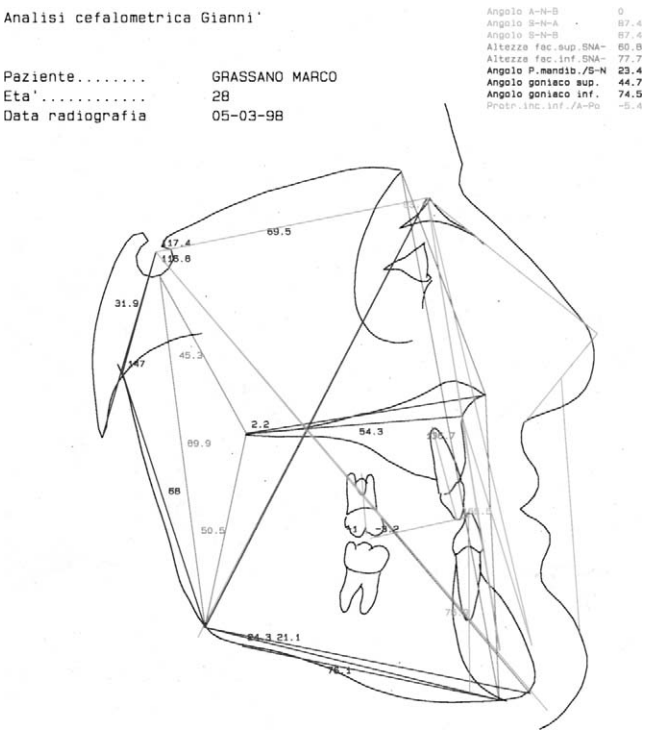

Figure 5. Cephalometric analysis.

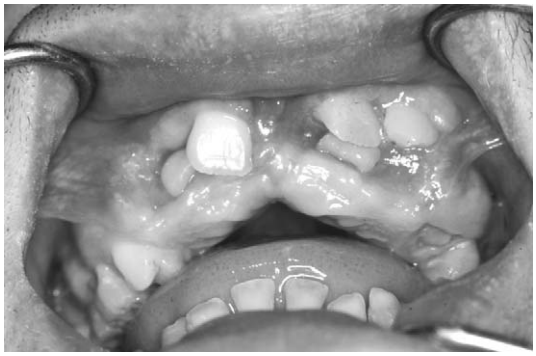

Figure 6. View after removal of the deciduous and supernumerary teeth, and surgical exposure of impacted teeth.

The patient had a skeletal Class III malocclusion and a balanced facial pattern. Intraoral examination revealed a mixed dentition, with poor oral hygiene and an Angle Class III molar relationship. Clinical observation showed missing maxillary and mandibular permanent teeth. However, intraoral examination showed many impacted teeth that did not erupt (Figures 1a–c and 2a–e). The oral inspection exhibited the following dental formula:

7 6 V IV III II I	I III IV V 6 7
7 6 5 4 III 2 1	1 2 III IV 5 6 7

The panoramic radiograph demonstrated the maxillary and mandibular impacted teeth, and the presence of supernumerary teeth in both regions. The radiograph analysis showed the presence of six supernumerary teeth, half in the maxilla, and the other half in the mandible (Figure 3).

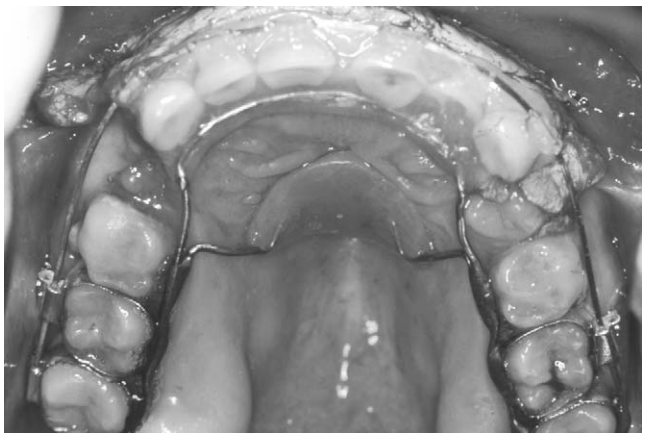

Figure 7. View of the intraoral anchorage.

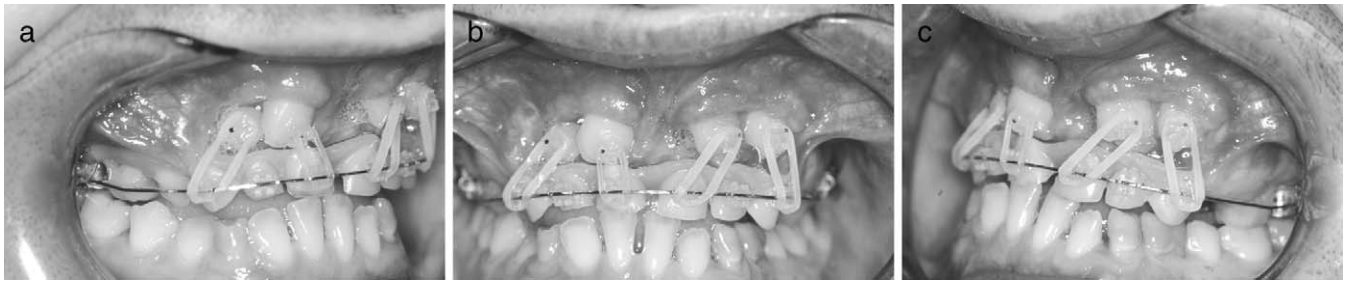

Figure 8. (a–c) Lateral and frontal intraoral view with elastic orthodontic traction.

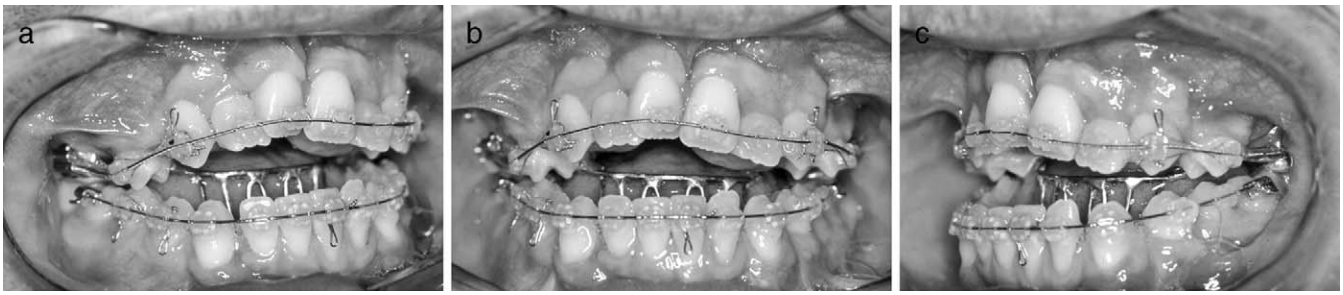

Figure 9. (a–c) Lateral and frontal intraoral view during the use of the lingual crib.

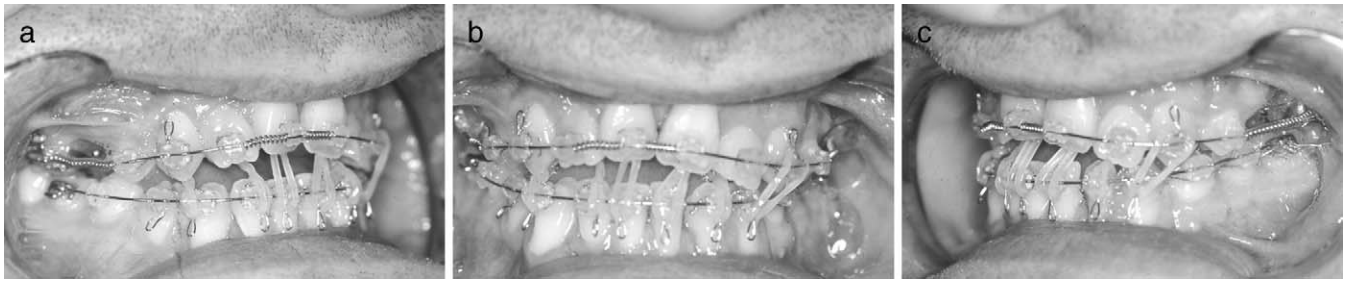

Figure 10. (a–c) Lateral and frontal intraoral view during the use of the elastic orthodontic traction.

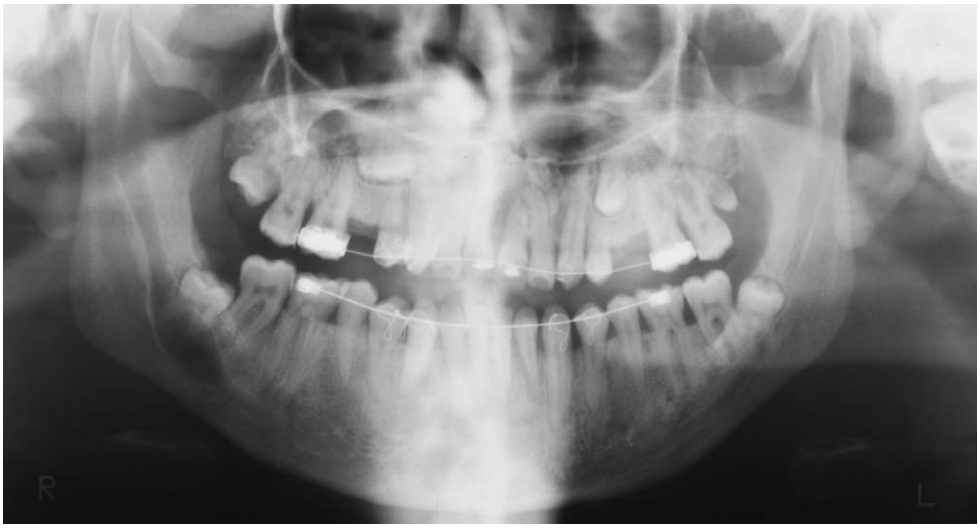

Figure 11. Panoramic x-ray at the end of the orthodontic treatment.

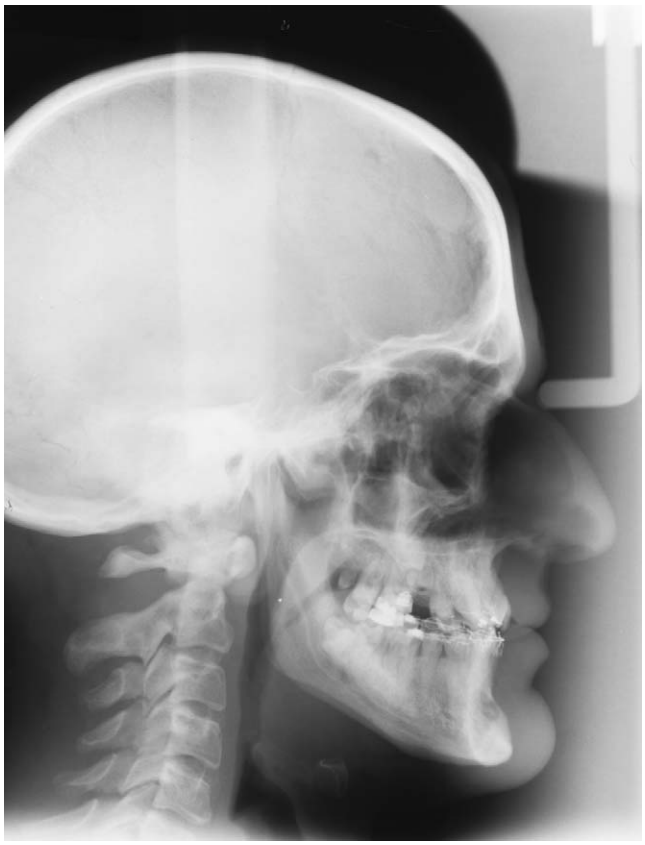

Figure 12. Laterolateral radiograph at the end of the orthodontic treatment.

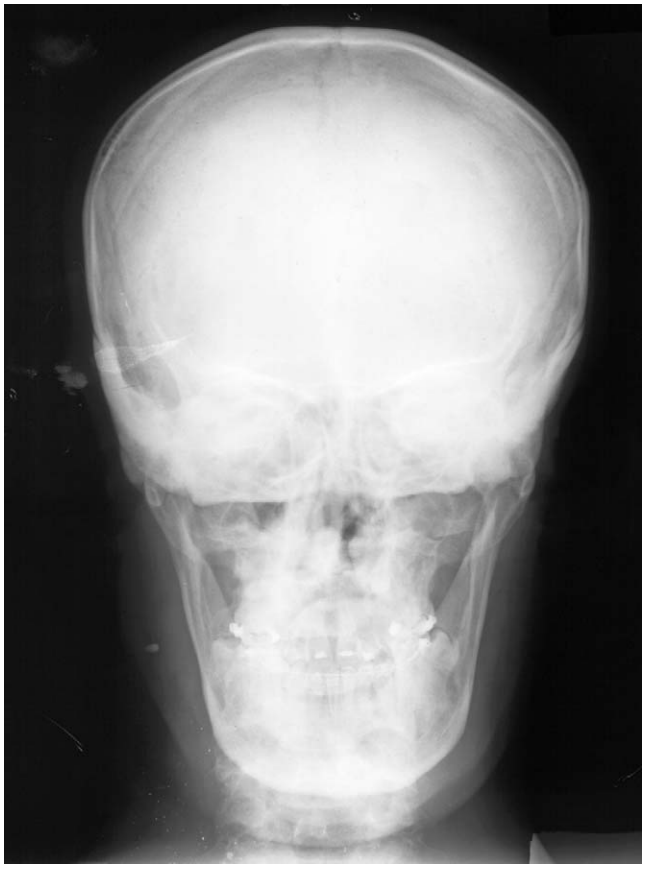

Figure 13. Posteroanterior radiograph at the end of the orthodontic treatment.

The craniofacial aspects were analyzed by means of a lateral radiograph. The cephalometric features confirmed the skeletal malocclusion. The SNA angle value was 87.4, SNB 87.4, and ANB 0. The upper gonial angle was 44.7 and the lower 74.5 (Figures 4 and 5).

Therefore, the patient was diagnosed with mandibular prognathism accompanied by multiple congenitally impacted and supernumerary teeth. The radiologic examination was ordered to analyze the skeletal morphology of the skull and the face, and to observe the dental development.

RESULTS

A rare case of CCD is presented that was not previously diagnosed, though the patient had a prolonged presence of the primary dentition and retention of permanent teeth. Orthodontic treatment was initiated with the following goals: complete dental eruption through surgical exposure of impacted teeth, correction of the anterior crossbite, and an increase in vertical dimension.

Because of the advanced age of the patient, the therapy was performed in different steps. The treatment plan consisted of extraction of the deciduous and supernumerary teeth and surgical exposure of permanent teeth (Figure 6). Afterwards, for beginning orthodontic therapy, the unique treatment option was the orthodontic traction of the impacted teeth using a powerful unity of anchorage (Figure 7).

Intraoral orthodontic traction was applied during the therapy to guide the teeth eruption to a correct position on the mouth (Figure 8a–c). This stage of the treatment was slow and required much cooperation from the patient.

When all teeth were engaged and a functional occlusal relationship had been established, the anterior crossbite was still present. A lingual crib was applied to restore a normal functionality of the tongue and to reduce the dental crossbite (Figure 9a–c). The attainment of the correct dental position was obtained with intraoral elastic traction that the patient had worn for many hours a day (Figure 10a–c). The panoramic radiograph, lateral, and posteroanterior skull radiograph exhibited the end of the treatment (Figures 11 through 13).

At the end of the orthodontic therapy, the upper in-

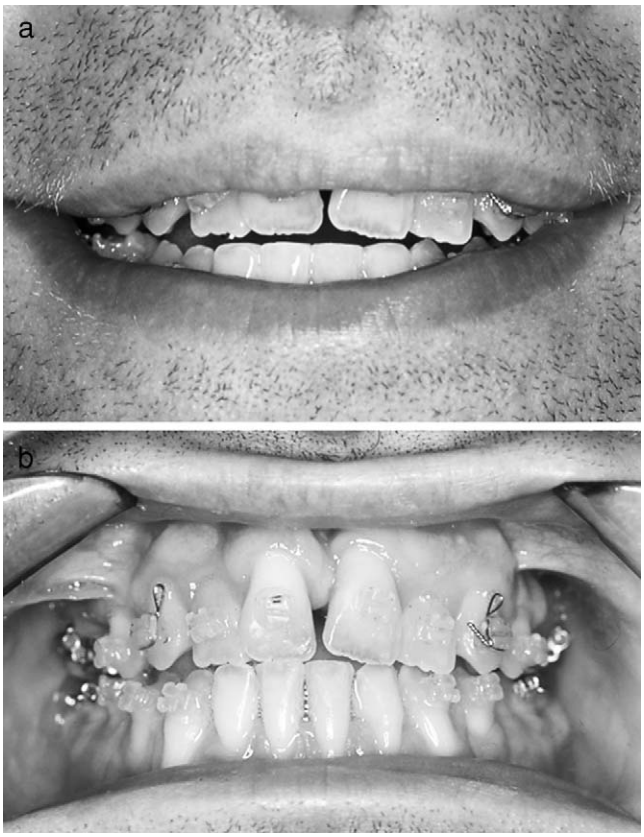

Figure 14. (a and b) View of the patient's smile and anterior teeth at the end of the orthodontic treatment

cisors eruption was not complete mineralize and not aesthetically acceptable (Figure 14a,b). Therefore, only for esthetic motivation a prosthetic treatment was performed on the six upper teeth (Figure 15a–c). The final stage of the orthodontic treatment was to stabilize the occlusal relationship. After active therapy, the patient used for one year the positioner, initially for a few hours a day and all night, and subsequently only during the night (Figure 16a–c).

After 1 year at the end of the orthodontic treatment it is possible to observe a functional occlusion and the aesthetic condition (Figures 17a–c and 18a–c).

DISCUSSION

This case demonstrates several characteristic anomalies in a patient with cleidocranial dysostosis. Because of the rarity of the condition, guidelines for the treatment of the CCD are rather sparse in the literature. Orthodontic treatment is usually indicated to direct the eruption of the malposed and often impacted teeth. The presented case illustrates also that the surgical, orthodontic, and prosthetic treatments were performed. Patients with cleidocranial dysostosis require a team approach with good cooperation and communication from the patient. Timing of the intervention is critical, and many surgeries might be required. It is very important also the collaboration of dentists with clinical geneticists to produce early diagnosis.

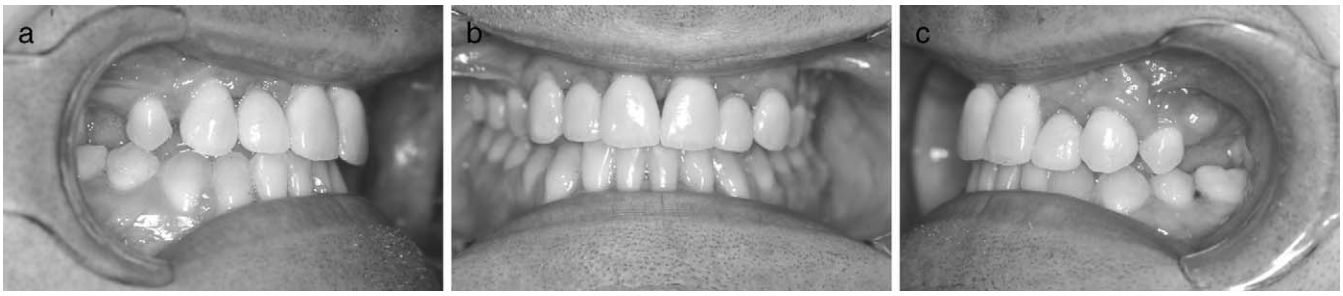

Figure 15. (a–c) Lateral and frontal intraoral view at the end of the prosthetic treatment.

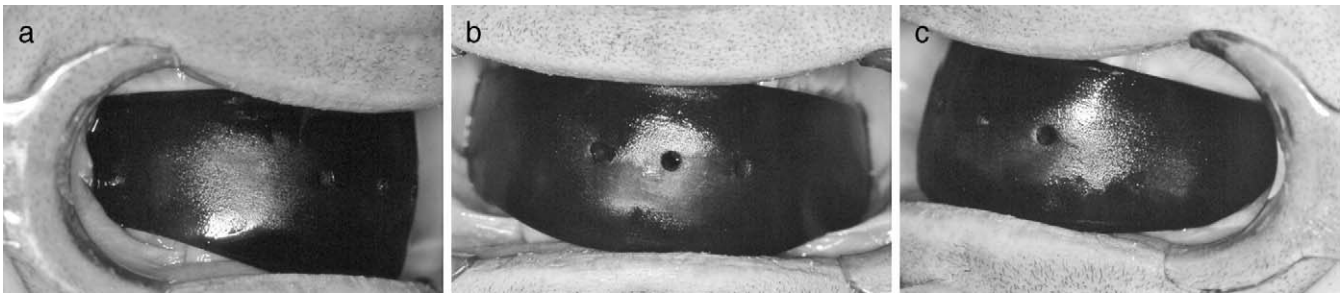

Figure 16. (a–c) Lateral and frontal intraoral view during the use of the positioner.

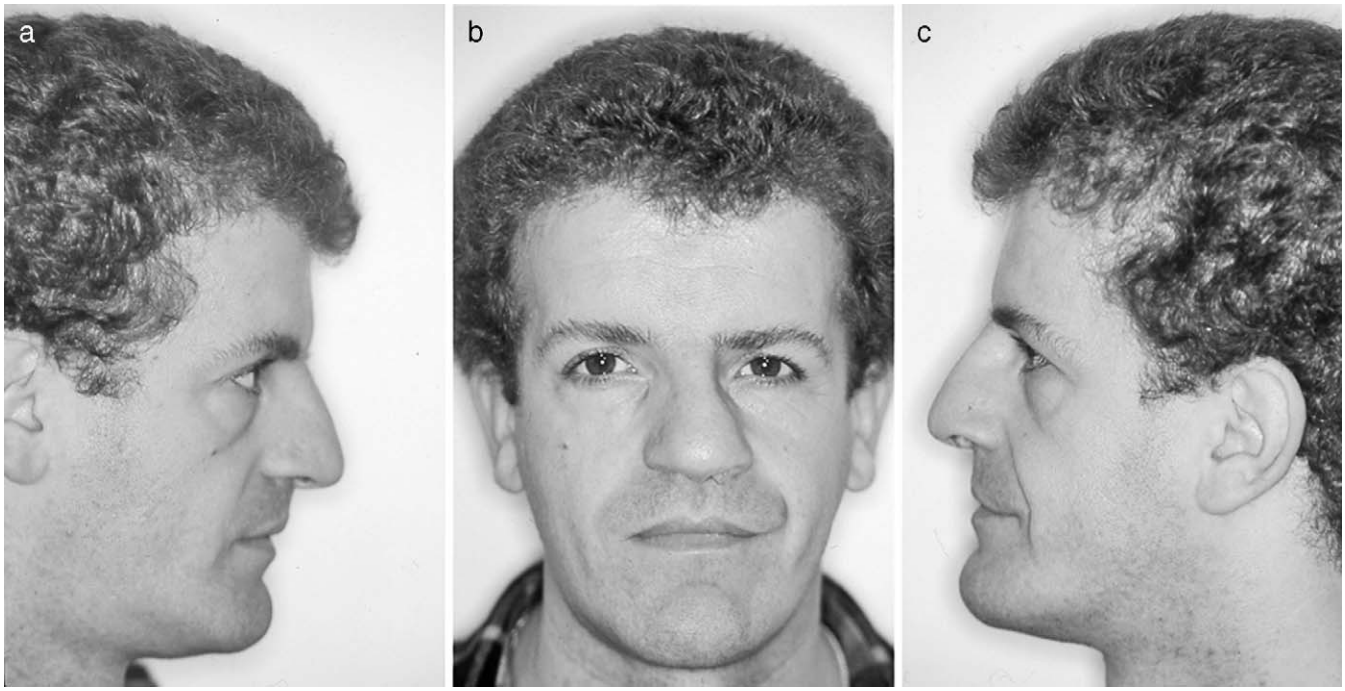

Figure 17. (a–c) Lateral and frontal view of the patient at the end of the treatment.

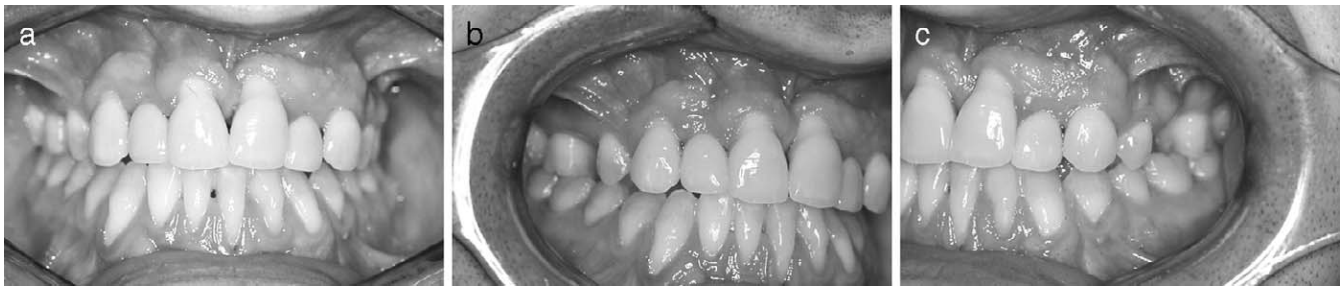

Figure 18. (a–c) Lateral and frontal intraoral view after 1 year of the end of the treatment.

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