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Management of Severely Atrophic Maxilla in Ectrodactyly Ectodermal Dysplasia-cleft Syndrome

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Background: Ectrodactyly ectodermal dysplasia-cleft syndrome is a rare genetic syndrome with an incidence of 1/90,000 live births, characterized by cleft lip and palate, severely hypoplastic maxilla, and hypodontia. Patients diagnosed with ectrodactyly ectodermal dysplasia-cleft syndrome suffer from a severely hypoplastic maxilla that is highly difficult to treat using traditional orthognathic methods. In this study, we propose using distraction osteogenesis to achieve a major advancement while maintaining good stability and minimal relapse. To our knowledge, this is the first description of patients with this syndrome treated using distraction osteogenesis.

Methods: Five patients diagnosed with ectrodactyly ectodermal dysplasia-cleft syndrome were included in the study. All patients had been operated on according to the well-established protocol of cleft lip and palate reconstruction before maxillary distraction osteogenesis. Hard and soft-tissue changes were evaluated by cone beam computed tomography and lateral cephalograms before distraction osteogenesis (T1), at the postdistraction point (T2) and after 1 year of follow-up (T3).

Results: Examination revealed marked maxillary advancement in all our patients with a significant mean difference in hard tissue parameters (condylion to A point = 18 mm; nasion-sella line to A point = 15.2 degrees) and a notable improvement in facial convexity (20.9 degrees). One year follow-up measurements demonstrated mild relapse rates of 6% in the horizontal plane.

Conclusions: We conclude that despite the challenging anatomic and physiological features of ectrodactyly ectodermal dysplasia-cleft patients, by enhancing current surgical techniques, there is promising potential for improved patient outcomes, achieving normognathic facial appearance with implant supported rehabilitation.
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INTRODUCTION

Ectrodactyly ectodermal dysplasia-cleft (EEC) syndrome is a rare genetic subtype of ectodermal dysplasia with an incidence of 1/90,000 live births, characterized by malformations in the extremities, defects in ectodermal structures, as well as cleft lip and palate.[1–3] Orofacial manifestations of EEC include a broad nasal base, abnormally long philtrum, indistinct vermilion border, and protuberant prolabium.[3] Moreover, in addition to cleft lip and palate, EEC manifests intraorally with a severely hypoplastic maxilla alongside underdeveloped thin alveolar ridges, hypodontia, malformed teeth, and loss of vertical dimension.[4,5] Primary repair of CLP in early childhood improves facial appearance, speech, and deglutition. However, it also causes malocclusion, secondary deformities, and maxillary growth impairment. Alongside, EEC comprises significant defect in the ectodermal structures including basal and alveolar bone that aggravate the deficiency.[6] Treatment of EEC patients is achieved by the coordinated efforts of a trained team of specialists including orthodontists, prosthodontists, and oral and maxillofacial surgeons.

To achieve normal facial proportions and intermaxillary relations, and improve the patients' quality of life, a marked maxillary advancement is required. Conventional

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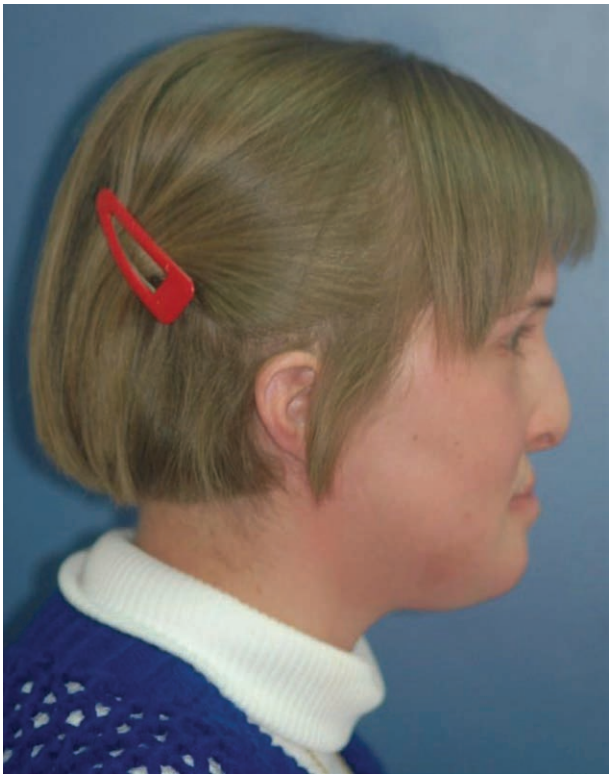

Fig. 1. Ten-year-old EEC patient. Lateral photograph showing maxillary hypoplasia and concave profile.

Le Fort I advancement with rigid fixation has been shown to be a reliable treatment modality for patients with mild deformities.[7–9] Regarding patients suffering from severe hypoplasia of the maxilla, reports advocate the use of distraction osteogenesis (DO) over the conventional Le-Fort I osteotomy.[10] Here, we present our experience with maxillary DO, specifically in EEC patients. We describe the challenging management of 5 individuals who had severely hypoplastic maxilla in both anteroposterior and vertical dimensions.

MATERIALS AND METHODS

For the current retrospective study, records of patients diagnosed with ectodermal dysplasia who presented to our department between 2002 and 2015 were reviewed. Five suitable patients were identified, all with a diagnosis of EEC syndrome (Fig. 1). All patients identified underwent maxillary advancement using DO and were included in this study, and none were excluded.

Patients included were operated on according to the well-established protocol of cleft lip and palate reconstruction, including lip and palate repair in infancy and early childhood and bone grafting to the cleft maxilla or alveolus between the ages of 8 and 11 years (Fig. 2). Before the DO procedure, radiographic imaging including panoramic x-rays, lateral cephalograms, and cone beam computed tomography were obtained, followed by an evaluation of hard- and soft-tissue parameters (Fig. 3). The presurgical evaluations, based on the aforementioned imaging

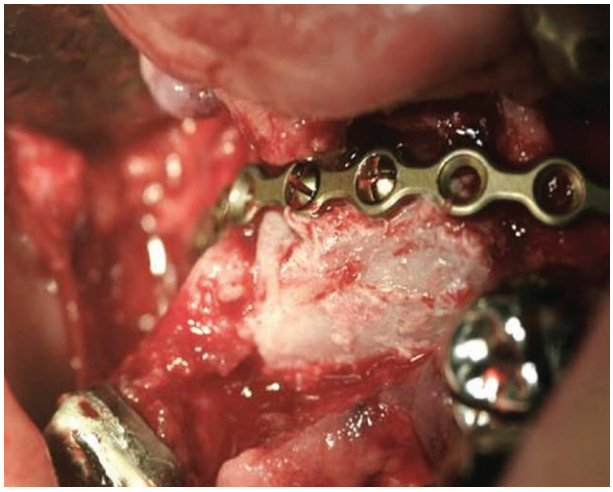

Fig. 2. The bone graft is positioned in the alveolar cleft maxilla and fixed with plate and screws.

methods, were carried out to assure adequate alveolar bone continuity, thus allowing a 1-piece maxilla osteotomy and mobilization (Fig. 4). According to the measurements performed on the clinical photographs, cephalograms and dental casts, the length and vector of elongation were planned, and distraction devices were ordered.

Surgical Technique

Generally, the maxillary distraction surgery was based on a Le-Fort I osteotomy according to Bell[11] and a fixation of 2 bilateral internal distractors (KLS-Martin, Jacksonville, Fla.). First, distraction appliances were adapted before the osteotomy with special attention to a forward and downward vector of elongation (Fig. 3). Second, due to a significant vertical loss of the maxillary bone, the maxillary osteotomy was confined between the infraorbital nerve superiorly and the dental roots (if existed) inside the atrophied alveolar crest inferiorly. The upper arms were then fixed to the zygomatic buttress, laterally and inferiorly to the infraorbital nerve and the lower arms to the alveolar crest. Due to the thin demineralized bone of EEC patients, it is highly important to ensure a proper anchorage of the intraoral devices, thus minimizing the risk of loosening hardware. The maxillary periosteum and mucosa were sutured leaving the bilateral activation rods in the maxillary vestibulum ready for activation (Fig. 5). Immediate postoperative radiographic images were obtained for all patients (Fig. 6).

A latency period of several days preceded the phase of active distraction, which is predetermined according to the planned elongation length, or usually around 3 weeks with a twice daily 0.5 mm elongation. We performed a slight overcorrection of 2–3 mm (Fig. 6). Throughout a 16-week consolidation period, weekly follow-ups were performed to prevent postoperative complications and to monitor patient weight and oral hygiene. Afterward, a second surgery was conducted for removal of both internal distractors. The patients then continued orthodontic treatment for postprocedural adjustments. The final step for establishing the patient's function and esthetics was

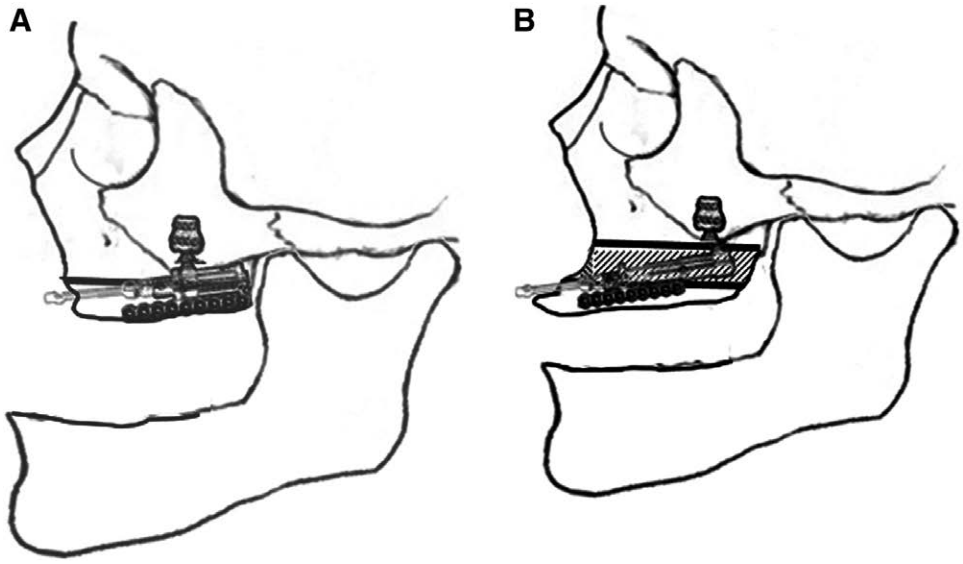

Fig. 3. Illustration demonstrating the Le Fort I osteotomy and distractor placement. A, Internal telescopic distraction device is fixed to the zygomatic buttress and the hypoplastic maxilla in a slight downward direction. B, Following the active distraction phase, note the newly formed bone represented by the shaded area.

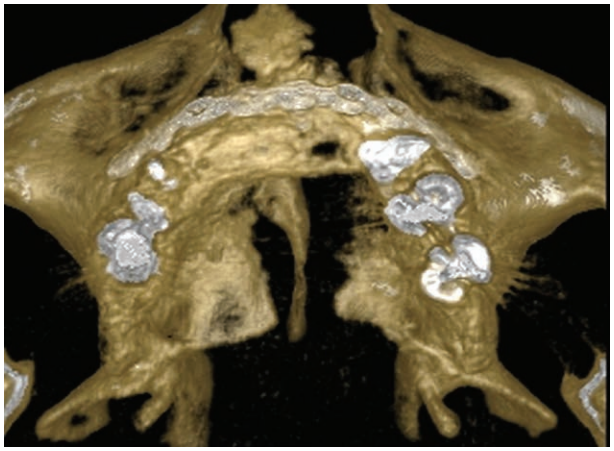

Fig. 4. The postsecondary alveolar cleft repair as shown in Figure 2. Three-dimensional cone beam computerized tomography illustrates a unified formation of bone in the premaxilla area following bone graft integration.

dental implant supported prosthetic rehabilitation, which was planned in accordance with postsurgical anatomic limitations (Fig. 7). The surgical sequence of treating EEC patients is presented in Table 1.

Demographic and treatment details of the 5 EEC patients who underwent DO in our department are summarized in Table 2.

Cephalometric Analysis

Hard- and soft-tissue evaluation was carried out by analyzing lateral cephalograms of all patients at 3 successive periods: Preoperative (T1), postdistraction (T2), and 1 year following removal of distraction devices (T3). Four skeletal and 3 soft-tissue reference points were chosen and measurements were recorded as follows: soft-tissue profile was evaluated by Glabella to Subnasale to Pogonion, G'SnPo (angle). The Nasion-Sella line to A point, SNA (angle), and Condylion to A point, CO-A were used to evaluate skeletal changes.

Ethical Approval

Rambam Health Care Campus Ethics Committee Approval No. 0423-09-RMB.

Patient Consent

Written consent of the patient was obtained for publication of the clinical photographs.

RESULTS

All EEC patients in this study demonstrated severe degree of maxillary hypoplasia, accompanied by cleft lip and palate and variable severity of oligodontia. During the period of the study, we treated 5 patients. Three of them were females (60%) and 2 males (40%) with a mean age of 16.8. Average latency and active distraction periods were 4 and 24 days, respectively, and the consolidation phase lasted for 20 weeks on average. Postoperative examination revealed marked maxillary advancement in all our patients (Figs. 7, 8). A Significant mean difference was observed with an increase of 18 mm and 15.2 degrees in maxillary length (Co-A) and SNA, respectively, and an additional improvement in facial convexity (20.9 degrees). Follow-up measurements were performed 12 months following removal of the distraction devices. As shown in Table 3, maxillary length was preserved with only 6% (average, 1.1 mm) posterior relapse. SNA angle was decreased by 2 degrees (13%) and facial convexity was reduced by 3.2 degrees (15.7%).

Two of our patients were monozygotic twins, who had been operated on simultaneously in our institution since

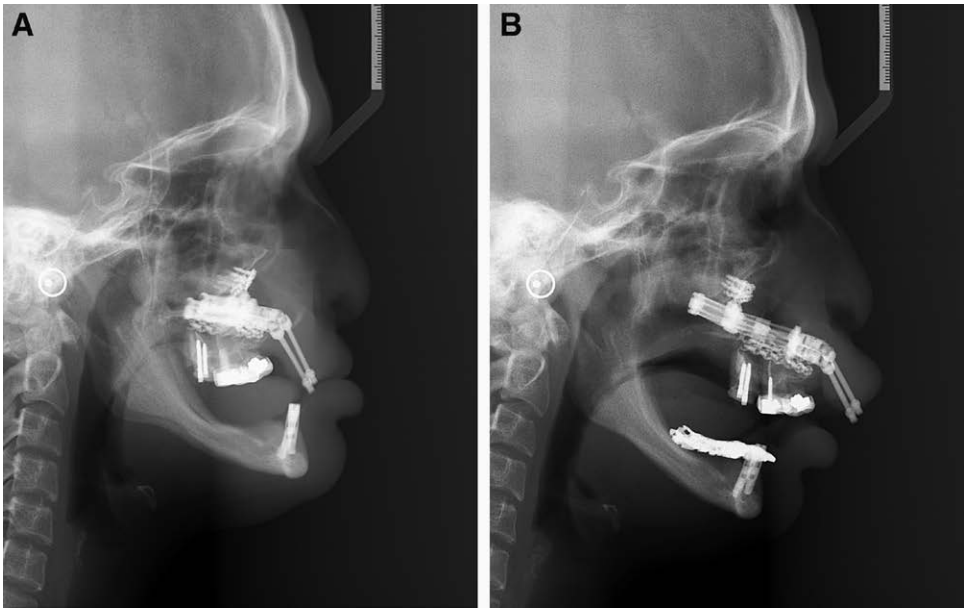

Fig. 5. Lateral cephalograms (A) of the hypoplastic retruded EEC patient following internal distraction device fixation. The devices were fixed to the zygomatic buttresses and maxillary process bilaterally. B, Following the activation phase, note the major maxillary advancement.

infancy. One of them experienced early closure of bone segments and an exposed right distractor during the active distraction phase, which required an immediate surgical intervention to allow mobilization of the maxilla and a new fixation of the intraoral distraction device.

DISCUSSION

Patients diagnosed with EEC syndrome often present with a severely hypoplastic maxilla with underdeveloped thin alveolar ridges, hypodontia, malformed teeth, and loss of vertical dimension. In this study, we suggest using DO to correct anterior–posterior deficiency resulting from the hypoplastic maxilla in EEC patients, allowing a major elongation and superior stability as compared with traditional orthognathic surgery. Five patients with rare ectodermal dysplasia and cleft lip and palate were treated using this modality in our institution. To our knowledge, this is the first description of EEC patients treated using DO.

A marked advancement of the maxilla and a slight vertical elongation was demonstrated in all patients. Significant improvement in facial profile resulting in a convex appearance was observed in all patients. All our patients were satisfied from the major improvement in facial appearance. The 12-month follow-up data showed stable results regarding skeletal advancement, with a mean horizontal relapse rate of 6% in maxillary length (Co-A) and 13% in SNA angle. This relapse was anticipated due to the major maxillary movement, and thus a slight over correction of 2–3 mm was performed.

These rates are lower than those reported in the literature among nonsyndromic CLP patients treated with orthognathic maxillary advancement in deficiencies of less than 8 mm, compared with our study, which presented a mean horizontal movement of 18 mm.[12,13] To our knowledge, there are only few reports in the literature elaborating the treatment of hypoplastic maxilla among EEC syndrome patients. Most of them used the traditional method of Le-Fort I osteotomy to achieve advancement of the maxilla.

Traditionally, advancement of the hypoplastic maxilla, regardless of etiology, is achieved with orthognathic surgery, using a conventional Le-Fort I osteotomy. Worsaae et al.[14] described the need for a Le-Fort I advancement in cases of oligodontia associated with EEC.[14] Posnick et al.[8] and Rachmiel et al.[9] have shown limitations of the Le-Fort I osteotomy to include difficult maxilla mobilization due to the formation of postsurgical scarring after cleft lip and palate repair, and high relapse rates. Moreover, large sagittal discrepancies between bony segments place patients at increased risk for velopharyngeal insufficiency.[15]

In previous works, application of the maxillary DO technique has demonstrated improved stability and greater maxillary advancement due to the slow lengthening and the concomitant bone and soft-tissue formation, as compared with a conventional Le-Fort I procedure.[6,16] Among the advantages of this technique are the ability to perform major maxillary movements, stability, less velopharyngeal insufficiency, and an additional elongation of adjacent soft tissues including muscles, nerves, and skin.

In the present study, internal distractors were used. The internal devices are almost invisible and causes less social burden to the patient and therefore allowed for longer consolidation period than the external devices that contribute to the postoperative stability.[6,7]

A highly important factor during active DO is controlling the vector of elongation to assure the desired positioning of the maxilla at the end of the DO process. Intermaxillary elastics are 1 option to use during the active

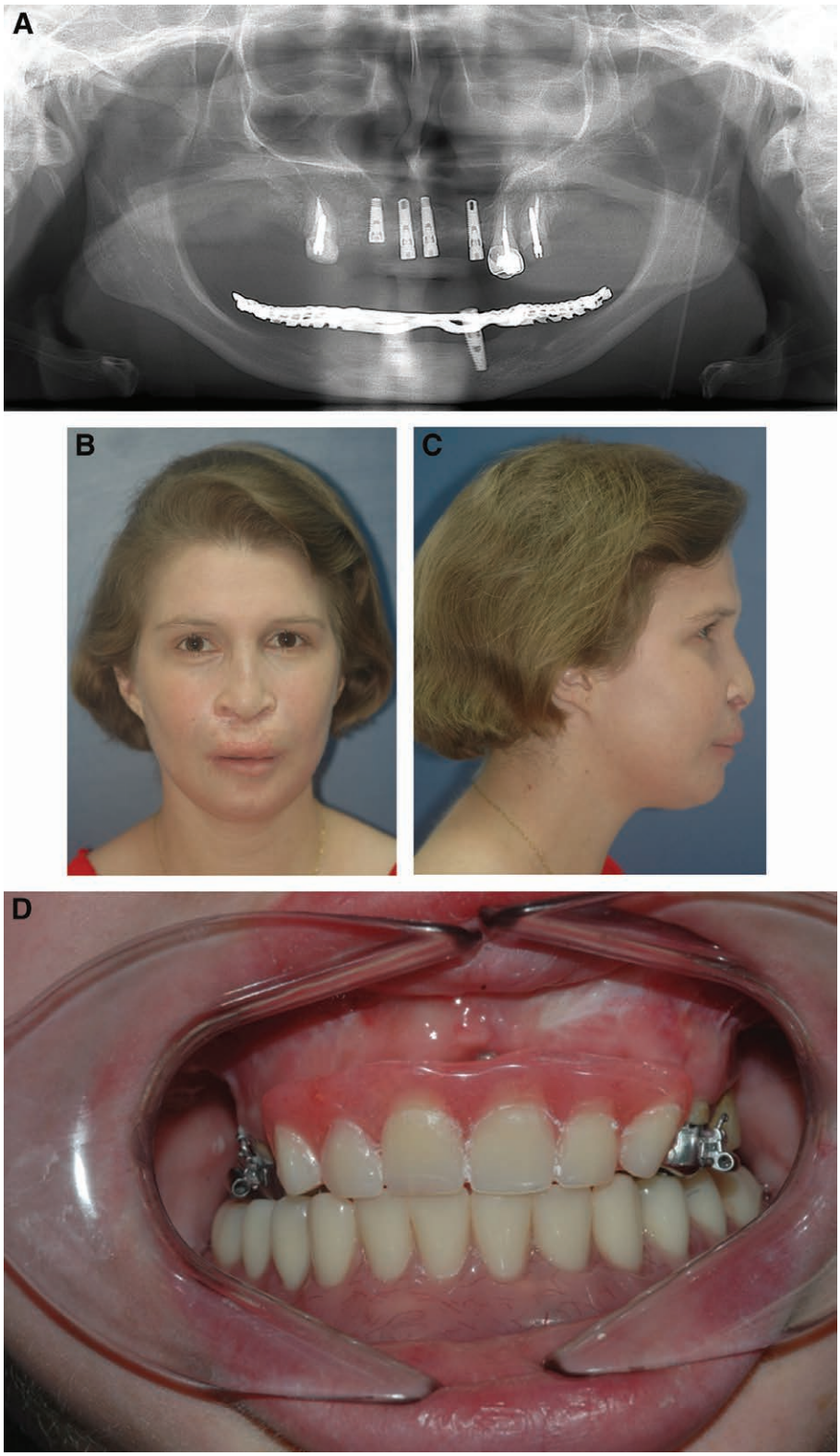

Fig. 6. Following dental rehabilitation. A, Panoramic radiograph showing the placement of dental implants in the maxilla. B, C, One-year postdistraction clinical photographs demonstrating normognathic facial appearance, notice the convex profile and the paranasal bony support. D, Prosthetic rehabilitation supported by the dental implants.

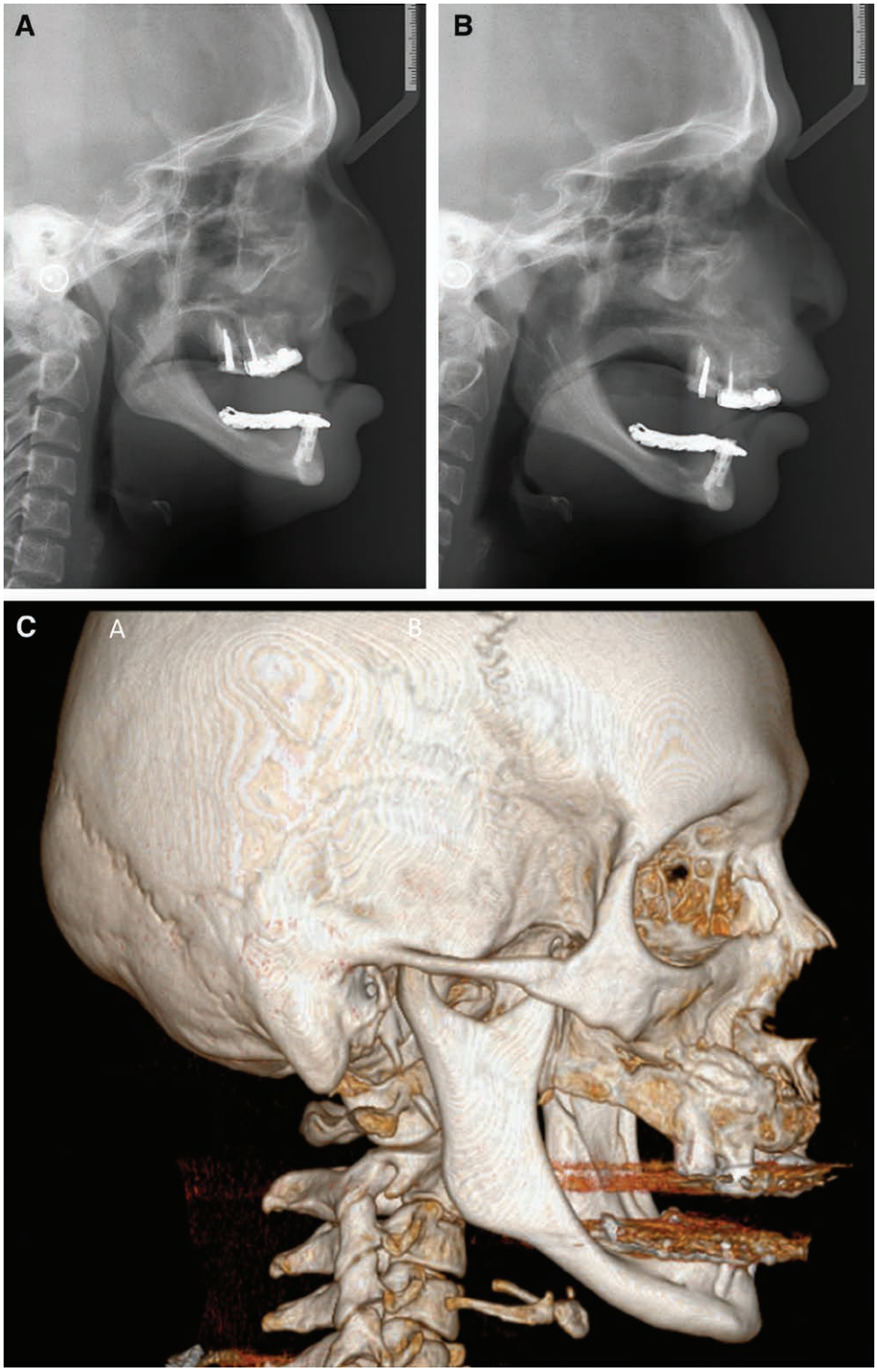

Fig. 7. Radiographs demonstrating the extensive maxillary movement. A, Preoperative lateral cephalogram demonstrates the retrognathic position of the maxilla resulting in a concave profile and edentulous aged appearance. B, The achieved facial convexity and sagittal maxillary advancement are presented in the postoperative lateral cephalogram. C, Three-dimensional computerized tomography demonstrates the newly formed maxillary bone resulting in normal intermaxillary relations.

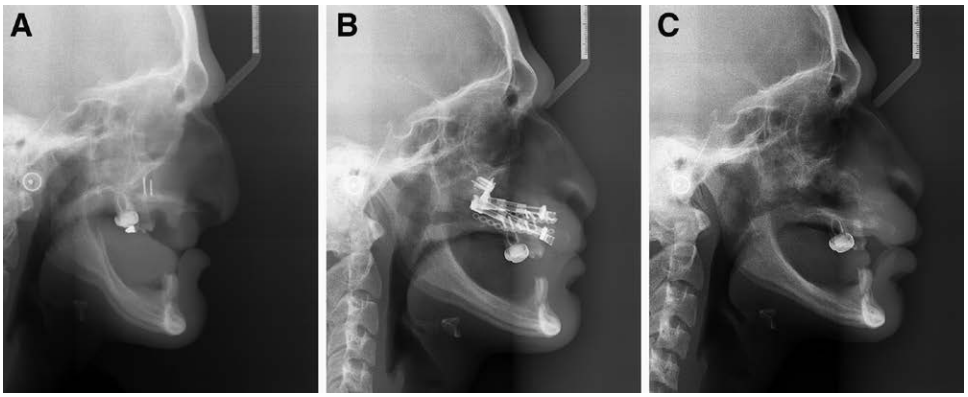

Fig. 8. Lateral cephalograms of another 17-year-old EEC patient suffering from severe hypoplastic maxilla (A) following internal distraction device fixation. The devices were fixed to the zygomatic buttresses and maxillary process bilaterally. B, Following the activation phase, note the major maxillary advancement. (C) One-year postdistraction stable results.

Table 1. Sequence of Surgical Treatments from Infancy to the Final Prosthetics

Infancy and Early Childhood	Age 8–11 y	Following Puberty	Final Treatment Adulthood	Final Treatment
Repair of cleft lip and palate	Secondary alveolar cleft repair using autogenous bone grafting	Maxillary advancement using DO	Additional maxillary and crestal bone augmentation as needed	Dental implant supported rehabilitation

Sequence of surgical treatments from infancy to the final prosthetics.

Table 2. Demographic and Treatment Details of the 5 EEC Patients Who Underwent Maxillary DO Advancement in Our Department

Patient	Age (y)	Sex	Cleft Diagnosis	DO Osteotomy	Latency Period (d)	Active Distraction (d)	Consolidation Period (mo)
1	19	F	Bilateral	Oblique	4	25	6
2	16	M	Bilateral	Oblique	3	24	4
3	16	M	Bilateral	Oblique	3	23	4
4	18	F	Bilateral	Horizontal	4	24	4
5	15	F	Bilateral	Horizontal	5	26	5

Demographic and treatment details of the 5 EEC patients who underwent maxillary DO advancement in our department.
F, female; M, male.

Table 3. Significant Mean Changes in All of the Measured Postoperative Parameters

	Preoperative T1 (n = 5)	Postoperative T2 (n = 5)	Delta T2–T1	Follow-up T3	Delta T2–T3	Relapse (%)
Co-A (maxillary length; mm)	73.3	91.3	18	90.2	1.1	6
SNA (degrees)	73	91.2	15.2	89.2	2	13.4
G'SnPo (facial convexity; degrees)	-15.9	4.9	20.9	1.7	3.2	15.7

A significant mean change in all the measured postoperative parameters.

distraction phase, yet in patients suffering from oligodontia this application is difficult. In our study, temporary anchorage devices were used as an alternative option in patients lacking the sufficient dentition for tooth borne elastics.[17–19]

In complicated DO procedures, such as in patients with EEC, the use of a computerized tomography designed stereolithographic model allows for presurgical adjustment of the devices and thus immediate and accurate fixation of the internal devices during surgery. Furthermore, the use of custom-made internal devices would allow a much more efficient and accurate advancement with better vector anticipation and thus superior results and fewer complications.

A hypoplastic maxilla has both functional and esthetic consequences in patients, which hamper quality of life and

preclude effective dental rehabilitation. We demonstrate that despite challenging anatomic features of EEC patients, there is promising potential for improved appearance, intermaxillary relations, and even dental rehabilitation in patients with ectodermal dysplasia. Further research is required to expand the existing body of knowledge regarding treating severe hypoplastic maxilla using DO.

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REFERENCES

1. Reed WB, Lopez DA, Landing B. Clinical spectrum of anhidrotic ectodermal dysplasia. *Arch Dermatol.* 1970;102:134–143.
2. Solomon LM, Keuer EJ. The ectodermal dysplasias. Problems of classification and some newer syndromes. *Arch Dermatol.* 1980;116:1295–1299.
3. Sharma D, Humar C, Bhalerao S, et al. Ectrodactyly, ectodermal dysplasia, cleft lip, and palate (EEC syndrome) with tetralogy of Fallot: a very rare combination. *Front Pediatr.* 2015;3:51.
4. Roelfsema NM, Cobben JM. The EEC syndrome: a literature study. *Clin Dysmorphol.* 1996;5:115–127.
5. Bondarets N, McDonald F. Analysis of the vertical facial form in patients with severe hypodontia. *Am J Phys Anthropol.* 2000;111:177–184.
6. Rachmiel A, Aizenbud D, Peled M. Long-term results in maxillary deficiency using intraoral devices. *Int J Oral Maxillofac Surg.* 2005;34:473–479.
7. Combs PD, Harshbarger RJ. Le Fort I maxillary advancement using distraction osteogenesis. *Semin Plast Surg.* 2014;28:193–198.
8. Posnick JC, Dagys AP. Skeletal stability and relapse patterns after Le Fort I maxillary osteotomy fixed with miniplates: the unilateral cleft lip and palate deformity. *Plast Reconstr Surg.* 1994;94:924–932.
9. Rachmiel A, Even-Almos M, Aizenbud D. Treatment of maxillary cleft palate: distraction osteogenesis vs. orthognathic surgery. *Ann Maxillofac Surg.* 2012;2:127–130.
10. Andersen K, Svenstrup M, Pedersen TK, et al. Stability after cleft maxillary distraction osteogenesis or conventional orthognathic surgery. *J Oral Maxillofac Res.* 2015;6:e2.
11. Bell WH. Le Fort I osteotomy for correction of maxillary deformities. *J Oral Surg.* 1975;33:412–426.
12. Polley JW, Figueroa AA. Rigid external distraction: its application in cleft maxillary deformities. *Plast Reconstr Surg.* 1998;102:1360–1372; discussion 1373.
13. Cheung LK, Chua HD, Hägg MB. Cleft maxillary distraction versus orthognathic surgery: clinical morbidities and surgical relapse. *Plast Reconstr Surg.* 2006;118:996–1008; discussion 1009.
14. Worsaae N, Jensen BN, Holm B, et al. Treatment of severe hypodontia-oligodontia—an interdisciplinary concept. *Int J Oral Maxillofac Surg.* 2007;36:473–480.
15. Harada K, Ishii Y, Ishii M, et al. Effect of maxillary distraction osteogenesis on velopharyngeal function: a pilot study. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod.* 2002;93:538–543.
16. Figueroa AA, Polley JW, Friede H, et al. Long-term skeletal stability after maxillary advancement with distraction osteogenesis using a rigid external distraction device in cleft maxillary deformities. *Plast Reconstr Surg.* 2004;114:1382–1392; discussion 1393.
17. Aizenbud D, Hazan-Molina H, Cohen M, et al. Combined orthodontic temporary anchorage devices and surgical management of the alveolar ridge augmentation using distraction osteogenesis. *J Oral Maxillofac Surg.* 2012;70:1815–1826.
18. Rachmiel A, Emodi O, Gutmacher Z, et al. Oral and dental restoration of wide alveolar cleft using distraction osteogenesis and temporary anchorage devices. *J Craniomaxillofac Surg.* 2013;41:728–734.
19. Shilo D, Emodi O, Aizenbud D, et al. Controlling the vector of distraction osteogenesis in the management of obstructive sleep apnea. *Ann Maxillofac Surg.* 2016;6:214–218.