Orthognathic Surgery in Cleidocranial Dysplasia

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Abstract: Optimal surgical correction of the craniofacial manifestations of cleidocranial dysplasia (CCD) has not been established due to the rarity of the condition. A 27-year-old female with CCD is presented. She underwent virtual surgical planning (VSP) followed by LeFort-I disimpaction, bone grafting, bilateral sagittal-split osteotomy, genioplasty, submental lipectomy, and targeted facial fat grafting. The patient necessitated 15-mm of vertical maxillary disimpaction centrally, stabilized with wide maxillary plates and interpositional allogenic fibula grafts. Six-month postoperative examination demonstrated improved appearance and functional symptoms. Skeletal relationships were normalized on computed tomography (CT) and there was minimal change between immediate and 6-month postoperative CT measurements, demonstrating a stable result. Orthognathic surgery used to establish dentofacial harmony in patients with CCD can test the extremes of single-stage facial skeletal expansion. Use of VSP, wide maxillary plates, and interpositional bone grafts can help optimize maxillary expansion and stability, while concurrent fat redistribution optimizes facial aesthetics.

Key Words: Class-III malocclusion, cleidocranial dysplasia, maxillary disimpaction, maxillary hypoplasia, orthognathic surgery

Cleidocranial dysplasia (CCD) is an autosomal dominant disease associated with abnormal bone regulation.[1,2] CCD is exceedingly rare, affecting 1 in 1,000,000, but has increased in public awareness in recent years.[3] Patients with CCD have a striking phenotype associated with an absent or hypoplastic clavicle, short stature, wormian bones, and widened pubic symphysis.[2] More notable are the craniofacial features which include a shortened and retruded midface, maxillomandibular malocclusion, hyperdontia and retained deciduous dentition, as well as cranial disproportion from delayed closure of the fontanels.[1,2]

Due to its rarity, there are few reports on the successful treatment of the craniofacial manifestations of CCD and no standard treatment approach has been established. Possible management options include orthognathic surgery or maxillary distraction.[4] Orthognathic surgery is single-stage procedure that theoretically poses a higher risk of relapse, while maxillary distraction may achieve larger advancements at the cost of two anesthesia events.[5] We report a unique approach combining virtual surgical planning (VSP), orthognathic surgery, and bone grafting for the successful treatment of a patient with craniofacial features of CCD.

CLINICAL REPORT

A 27-year-old female presented with a chief complaint of facial dysmorphism, dyspnea, snoring, trouble eating, and intermittent jaw pain. She was previously diagnosed with CCD at an outside institution. Prior orthodontia included several years of braces. The patient has no significant family history of this disease. On exam she demonstrated significantly shortened midfacial height, a concave facial profile with hollowness of the perialar regions and class II malocclusion, as well as mild temporomandibular joint tenderness (Fig. 1). The patient also exhibited anterior crossbite, a narrow maxilla, and posterior crossbite with offset of maxillary and mandibular midlines.

The patient was scheduled for LeFort-I osteotomy, bilateral sagittal split osteotomy (BSSO), osseous sliding genioplasty, submental lipectomy, and fat grafting to key areas of the face. VSP was used to plan the degree of maxillary disimpaction, mandibular correction, and repositioning of the genioplasty segment (Fig. 2). Intraoperatively, BSSOs were performed first and an intermediate occlusal splint was used to guide plating of the mandible. Next, LeFort-I osteotomy was performed, noting the underdeveloped maxillary sinuses. Due to the patient's pathologically short maxilla, disimpaction at the piriform was performed slightly beyond what was planned virtually: 15-mm at the central incisors and 13.5 mm at the canines. The patient's dental show at rest was used to fine-tune the level of disimpaction on-table. Standard midface L-plates were placed laterally, while wide maxillary plates were used for fixation

FIGURE 1. Preoperative and postoperative photos, virtual surgical plan, and intraoperative stabilization. A) preoperative anterior view photograph; B) preoperative profile view photograph; C) fibular bone allograft segments and wide plates placed at the nasomaxillary buttresses; D) stabilization of the BSSO with 1.0 mm plate and two mono-cortical screws on either segment bilaterally; E) Genioplasty performed with 5 mm of down-fracture and 2 mm of advancement, stabilized with a prebent titanium plate. F) postoperative anterior view photograph; G) postoperative profile view photograph.

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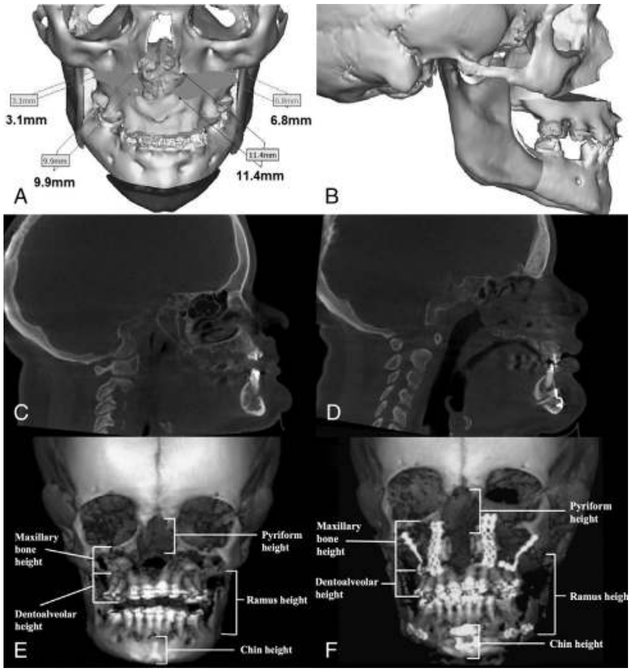

FIGURE 2. Virtual surgical plan and preoperative versus postoperative CBCT and 3D reconstructions. A) virtual surgical plan bird's eye view; B) virtual surgical plan lateral view; C) preoperative lateral CBCT demonstrating severely limited maxillary height and projection; D) postoperative lateral CBCT demonstrating improved maxillary and mandibular dimensions; F) preoperative 3D reconstruction anterior view; G) postoperative 3D reconstruction anterior view.

at the pyriform with two 14-mm segments of allogenic fibula predrilled and screwed interpositionally for added stability (Fig. 1). A subsequent osseous sliding genioplasty augmentation was performed with 5-mm of advancement and 1-mm of disimpaction to improve the labiomental sulcus and chin position on profile view. Allogenic demineralized bone matrix was used to fill in the gap above the genioplasty segment and at the BSSO plating sites. In addition, to correct the patient's submental and cervical lipodystrophy, submental suction assisted lipectomy (SAL) was performed, removing ~70 cc of lipoaspirate. Finally, fat from the abdomen was harvested, prepared, and injected into the midface, labiomental crease and lateral chin area.

Cone-beam computed-tomography (CBCT) and clinical photographs were obtained preoperatively and on postoperative day 1 and month 6 (Fig. 2). Cephalometric analysis was performed using CBCT scans analyzed in Materialise Mimics Medical v21.0 (Leuven, Belgium). The Frankfort horizontal (FH), defined by Orbitale and the superior aspect of the external auditory canal bilateral, was used as an axial reference plane. A plane through Nasion, Crista galli, and Cribriform plate perpendicular to FH was used to create a Midsagittal plane (MS). The palatal plane included the anterior nasal spine (ANS) and posterior nasal spine (PNS) drawn perpendicular to the midsagittal plane. All vertical distances were measured plumb/perpendicular to FH and parallel to MS. Standard cephalometric angles were measured. In addition, maxillary bone height was measured as the vertical distance from Orbitale to the palatal plane on both sides. Dentoalveolar height was measured as the vertical distance from ANS to the occlusal plane. Total maxillary height was computed by adding maxillary bone height to dentoalveolar length. Mandibular ramus height was measured as the vertical distance from Condylion to Gonion. Chin height was measured as the vertical distance from B-point to Menton. Chin point deviation was defined as the horizontal distance from Menton to MS.[13] Pyriform height was the vertical height of the Pyriform aperture at its tallest point, measured bilaterally.

RESULTS

Postoperatively, the patient recovered from the procedure well without complications over a 10-month follow-up period. Clinically, the patient demonstrated significantly improved facial profile, skeletal harmony, dental show, occlusion, and jaw function. Caliper measurements performed on-table immediately postop from the left medial canthus to the central incisor edge, left medial canthus to the left canine edge, and right medial canthus to the right canine edge were 62, 54.5, and 58 mm, respectively; compared to 46.5, 41, and 44 mm at the start of the case, denoting significant maxillary movement.

Cephalometrically, there was elimination of the occlusal cant, maxillary midline offset, mandibular midline offset, and chin point deviation (Supplemental Table 1, http://links.lww.com/SCS/C9). Increases in pyriform height, maxillary bone height, total maxillary height, ramus height, and chin height were 7.77, 10.99 (doubled from preop), 11.48, 2.02, and 2.90 mm, respectively. Dentoalveolar height did not change significantly (+0.49 mm). Additionally, there were also large increases in occlusal plane angle (+19.10°), facial angle (+9.87°), gonial angle (+8.00°), SNA (+4.85°), and SNB (+4.51°). There were minimal changes in the skeletal position from postoperative day 1 to 6-month (Supplemental Table 1, http://links.lww.com/SCS/C9). These findings demonstrate that the orthognathic movements and technique described improved facial balance with stable results. By elongating the facial skeleton and performing SAL, the patient's perceived facial and cervical obesity was much improved as well, despite the patient gaining 2-kg in weight (height 150 cm, BMI: 35.3 preop, 36.2 six-months postop).

DISCUSSION

The outcomes presented in this report demonstrate the benefits of utilizing a multi-disciplinary approach involving surgeon coordination with the orthodontist and VSP to optimally deliver orthognathic surgery in CCD. In combination with fat redistribution, this single-stage procedure can provide tremendous improvement in facial aesthetics and function. VSP used in this case allowed for improved intraoperative efficiency and technical accuracy which contributed to the patient's postsurgical outcome.[6]

This report is one of the first using interpositional bone grafts to help stabilize a significant maxillary disimpaction in CCD. This significant immediate skeletal expansion utilizing bone allograft and wider plates achieved stable outcomes without the need for maxillary distraction or a second visit to the operating room for hardware removal. Bone grafting can contribute to increased rates of relapse secondary to decreased bone support after infection or resorption.[5] However, with the wider plates and corticocancellous allograft used, relapse was not observed over the follow-up period. Interpositional bone grafts in orthognathic surgery have been shown to contribute to the stability of movements.[5]

Possible alternatives for the treatment of CCD include orthognathic surgery without bone grafting or maxillary distraction.[7] Maxillary advancement through LeFort-I osteotomy has resulted in improved occlusal relationship with stable and satisfactory postoperative results.[8–10] Single-stage reconstruction with orthognathic surgery is generally the preferred technique in skeletally mature patients that require mild to moderate advancement.[5] The use of maxillary distraction is advocated for defects that require larger advancement (>1 cm) in order to avoid the risk of relapse associated with orthognathic surgery and has also resulted in satisfactory postoperative outcomes.[5] Maxillary distraction is also appropriate for expansion of the maxilla prior to skeletal maturity. However, it necessitates additional procedures and does not circumvent the need for orthognathic surgery to correct final occlusion.

To completely improve the patient's facial balance and nasal breathing, the next step in her management is septorhinoplasty. Using an open approach, the patient will undergo septoplasty, caudal septal extension grafting to derotate the nose and add nasal length and projection, as well as dorsal recontouring and radix augmentation to optimize her facial profile. This study was limited by providing a single case with only 10-months follow-up. As an extremely rare condition, large case series at a single institution are not feasible. Further study may include additional case examples across multiple institutions with longer follow-up.

CONCLUSION

Orthognathic surgery used to establish dentofacial harmony in patients with CCD can test the extremes of single-stage facial skeletal expansion. Use of VSP, wide maxillary plates, and interpositional bone grafts can help optimize maxillary expansion and stability, while concurrent fat redistribution optimizes facial aesthetics.

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