A Subcranial Le Fort III Advancement With Distraction Osteogenesis as a Clinical Strategy to Approach Pycnodysostosis With Midface Retrusion and Exorbitism

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Abstract: Pycnodysostosis is a rare autosomal recessive skeletal disorder involving a constellation of craniofacial manifestations including midface retrusion. We report the case of a 13-year-old girl with pycnodysostosis who presented with exorbitism, midface retrusion, malocclusion, and obstructive sleep apnea. Here, we describe the successful use of subcranial Le Fort III advancement using distraction osteogenesis with internal Kawamoto distracters. After a latency of 5 days, distraction for 10 days, and consolidation for 12 weeks, her midface was advanced by 10 mm with slight overcorrection at the occlusion level. At 2 years postoperatively, the patient had complete remission of her sleep apnea, resolution of her exorbitism, and amelioration of her class III malocclusion to class I.

Key Words: Pycnodysostosis, distraction osteogenesis, midface retrusion, exorbitism, subcranial Le Fort III advancement

Pycnodysostosis (OMIM 265800) is a rare autosomal recessive skeletal dysostosis resulting from a mutation in the gene encoding cathepsin K, a lysosomal cysteine proteinase that is integral for normal osteoclastic bone resorption and remodeling. The disease is characterized by generalized bony sclerosis, bone fragility, short stature, dysplastic clavicles, and numerous craniofacial deformities, including dolichocephaly, persistent patency of 1 or more of the cranial fontanelles, midface retrusion or hypoplasia, micrognathia, malocclusion, and delayed dental eruption. Moreover, osteomyelitis of the mandible and recurrent pathologic fractures of the long bones are characteristic complications of this disease, owing to the impaired vascularity of the bone. Diagnosis of this disease is usually made by the association of the patient’s history and clinical findings, which is then substantiated by gene studies identifying the cathepsin K gene mutation.

Although pycnodysostosis has a characteristic phenotype and distinct genetic mutation, the rarity of the disease makes it a challenging diagnosis to identify. As such, there are very few reports of surgical management of these patients. Long-term stability of the midface is one of the primary concerns among craniofacial surgeons; and although midface advancement would serve as an effective solution to this problem, there has been apprehension surrounding the technique’s potential morbidity when used on bone that is already dysvascular, fragile, and considered unlikely to consolidate appropriately. Thus, craniofacial surgeons may turn away from midface advancement as a surgical option, and consequently, what is considered a less morbid orthognatic surgical approach is often used instead. Unfortunately, orthognatic techniques do not address the clinical exorbitism and midface retrusion, which are critical features of this disease; and as an unfortunate consequence, suboptimal aesthetics and postoperative function often result. Therefore, aside from the speculation of potential morbidity, midface advancement with distraction osteogenesis would seem to be the optimal surgical approach for midface retrusion and exorbitism associated with this particular disease.

CLINICAL REPORT

A 13-year-old girl presented to our clinic with obstructive sleep apnea, inability to spontaneously close her eyelids, and eye irritation and pain. She had a clinical history significant for craniosynostosis at the age of 15 months, surgically corrected with strip craniectomy at the coronal suture region, as well as multiple fractures since her first years of life. On physical examination, she was noted to have short stature, midface retrusion, class III malocclusion with dental crowding and posterior crossbite and exorbitism, as well as acroosteolysis.

Reformatted computed tomographic imaging showed patency of coronal and lambdoid cranial sutures, retrusion of the midface, including the zygomatic-orbital regions, and obtuse angle of the mandible. Plain radiographs showed a small airway pharyngeal space. Genetic evaluation revealed a p.Arg241X mutation in the cathepsin K gene, confirming the diagnosis of pycnodysostosis as previously reported by our group.

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Given the risk of infection and/or bone nonunion after an acute subcranial Le Fort III advancement with bone grafting, we decided to conduct a subcranial Le Fort III advancement with distraction osteogenesis. The procedure was performed through a coronal incision. Bilateral osteotomies were made in the anterior zygomatic region, lateral orbital wall, medial orbital wall and orbital floor and ptgeromaxillary buttresses, and at frontalosanal region. Rowe disimpaction forceps were used to down-fracture the face and to promote the final craniofacial disjunction. Bilateral univector internal Kawamoto distracters were fixed to the zygomatic body anterior to the osteotomy site and temporal bone, behind the zygomatic root and above the external auditory canal. Both devices were tested, and 2 drains were placed in the coronal region before closure.

The face was brought forward 10 mm, and a slight overcorrection at her occlusion level was performed by the standard protocol of distraction osteogenesis. The latency phase was 5 days, and consolidation phase was 12 weeks. At 2 years postoperatively, the patient had complete reposition of her sleep apnea, resolution of her exorbitism, and marked improvement of her class III malocclusion, which converted to a class II malocclusion immediately postoperatively. Her occlusion was stabilized at class I with an orthodontic appliance at 2 years after surgery. She maintains a posterior crossbite that will be corrected when her growth is complete (Figs. 1–3).

**DISCUSSION**

The poor bone vascularity and bone sclerosis found in pycnodysostosis have been cited as risk factors for the development of osteomyelitis of the maxillofacial skeleton and pathologic facial fractures. Moreover, these factors are thought to contribute to an increased risk for perioperative complications. Indeed, osteomyelitis-induced mandible defects have been repaired with titanium plates and/or nonvascularized iliac crest grafts. However, as failure occurred, vascularized iliac bone grafts and free fibular osteocutaneous flaps have been adopted as more effective alternatives.

Therefore, the tendency among surgeons has been to use conservative management techniques for craniofacial abnormalities associated with pycnodysostosis. This notion is reflected in the literature as our extensive English search (Medline and Embase databases) revealed a general paucity of reports that describe surgical methods addressing this disease. Most of what we encountered regarded the use of orthognatic techniques (Table 1).

The major advantage in undertaking orthognatic correction in patients with pycnodysostosis is the ability to achieve an adequate correction of the malocclusion without the morbidity of a major midface advancement. However, orthognatic surgery does not treat the exorbitism, and even high Le Fort I advancements may also be inadequate when used to address inferior orbital rim retrusion. Moreover, orthognatic surgery is not without its own set of complications. In particular, transection of bilateral inferior alveolar nerves has been reported after the creation of bilateral mandibular osteotomies, contributing factors to this complication being the sclerotic nature of the bone.

Della Marca et al reported airway augmentation using adenotonsillectomy and uvulo-palato-pharingoplasty in a 5-year-old patient with pycnodysostosis and obstructive sleep apnea. Although effective in treating the sleep apnea and avoiding a highly morbid tracheostomy, this procedure was unable to address the patient’s facial dysmorphisms. In contrast, a midface advancement with distraction osteogenesis, commonly used in syndromic craniosynostotic children with obstructive sleep apnea, can correct both sleep apnea and midface retrusion without jeopardizing speech function.

Other techniques involving bone grafting have also been described. Teissier et al used autologous rib grafting to advance the mandible and increase the volume of the pharyngeal port in a young patient not unlike ours. Although they achieved an excellent result in the end, there are persistent concerns that the lack of blood supply of the sclerotic bone in pycnodysostosis still put the patient at undue risk for osteomyelitis among other associated risks.

Midface advancement either with a subcranial Le Fort III or a monobloc advancement can simultaneously improve obstructive sleep apnea secondary to posterior pharyngeal soft tissue crowding and the exorbitism without requiring the morbidity of autologous bone grafting. Our group has extensive experience in subcranial Le Fort III and gradual monobloc advancement in treating craniofacial dysostosis with similar clinical presentation, and for this particular case we decided on the former approach. Interestingly, neither operative technique has been described as a primary treatment for pycnodysostosis, although some have considered distraction osteogenesis a form of adjuvant care for these patients, regardless
FIGURE 3. Preoperative lateral cephalograms showing the midface retrusion and the superior-inferior dental relation (left). Immediate postoperative lateral cephalograms taken at day 1 postoperatively, during the latency phase, and at 5 days before the beginning of activation phase (center). This radiographic imaging showed the positioning of the distracters related to the cranial base and the vectors of the distraction osteogenesis. The distracters were placed parallel to the other one. Postoperative lateral cephalograms taken at the last day of activation phase showed the amount of movement obtained, in addition to the dimensions of the posterior airway gained (right).

TABLE 1. Summary of Studies Containing Surgical Approach to Patients with Pycnodysostosis With Craniofacial Abnormalities According to our Literature Review7,12,14,16,17

<table>
<thead>
<tr>
<th>Author (y)</th>
<th>Patient Demographics (Age, Sex, and Ethnicity)</th>
<th>Craniodentofacial and Extremity</th>
<th>Clinical Findings</th>
<th>Radiographic Findings</th>
<th>Craniofacial Surgical Technique</th>
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<tbody>
<tr>
<td>Ilankovan and Moos (1990)</td>
<td>17 y, F, Pakistani</td>
<td>Frontal bossing, bilateral proptosis of the eyes, gross midface hypoplasia, beaked nose, small mandible, marked retroglossa, and class III malocclusion</td>
<td>X-ray: skull—sclerosis of skull base, patent frontotemporal and parietooccipital sutures; face—rudimentary maxillary sinuses and obtuse gonial angles of mandible; hands—short terminal phalanges and resorption of the terminal phalanges</td>
<td>Advancement of maxilla, mandible with inverted osteotomy, and genioplasty with bone grafting</td>
<td></td>
</tr>
<tr>
<td>Norholt et al (2004)</td>
<td>15 y, F, Danish</td>
<td>Midfacial hypoplasia, exophthalmos, blue sclera, and class III malocclusion</td>
<td>Lateral cephalogram: maxillary hypoplasia and obtuse mandibular angle</td>
<td>Bimaxillary distraction osteogenesis with rigid external device</td>
<td></td>
</tr>
<tr>
<td>Teissier et al (2009)</td>
<td>3.5 y, M, French</td>
<td>Severe upper airway obstruction with apnea, retrognathia, glossoptosis, beaked nose, high forehead, large fontanelles, and sagittal sutures</td>
<td>CT: hypoplastic mandible and maxilla, flattened mandibular angles, and bilateral exophthalmia</td>
<td>Costochondral rib grafting to enlarge pharyngeal airway</td>
<td></td>
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<td>Varol et al (2011)</td>
<td>33 y, F, Turkish</td>
<td>Midfacial hypoplasia, slight retrognathia, exophthalmos, and frontal bossing</td>
<td>Cephalometric analysis: reduced facial height and absence of gonial angle</td>
<td>Bimaxillary distraction osteogenesis with rigid external device</td>
<td></td>
</tr>
<tr>
<td>Hernandez-Alfaró et al (2011)</td>
<td>18 y, F, Spanish</td>
<td>Brachycephaly, open cranial sutures and fontanelle, maxillary and mandibular hypoplasia, rhinomelagly, and class II malocclusion</td>
<td>Cone beam CT scan: distorted facial architecture with abnormally small facial bones</td>
<td>Mandibular advancement with bilateral sagittal split osteotomy, maxillary advancement with Le Fort I osteotomy, and vertical sagittal augmentation genioplasty with synthetic bone graft</td>
<td></td>
</tr>
<tr>
<td>Raposo-Amaral et al (current study)</td>
<td>13 y, F, Brazilian</td>
<td>Midfacial hypoplasia, exorbitism, obstructive sleep apnea, and class III malocclusion</td>
<td>CT: patency of coronal and lambdoid cranial sutures and obtuse angle of the mandible</td>
<td>Subcranial Le Fort III advancement with internal distraction osteogenesis</td>
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F indicates female; M, male; CT, computed tomography.
of the type of osteotomy used. Reviewing the literature, we found little information regarding recommended periods for latency, activation, and consolidation for this particular disease; and so we adopted a traditional distraction osteogenesis protocol, reasoning that this would allow gradual bone regeneration and sufficient bone stability. Indeed, 2 years after surgery, the patient had sufficient stability of the midface, complete remission of the sleep apnea, and resolution of symptoms related to exorbitism such as corneal exposure, ocular redness, and ocular pain.

For this particular disease, we feel that subcranial Le Fort III advancement is able to correct more craniofacial dysmorphisms of pycnodysostosis than orthognatic procedures. Moreover, it produces a more stable result with less surgical morbidity in comparison to acute advancements involving bone grafts and rigid fixation.

In conclusion, this is the first report on the successful use of subcranial Le Fort III midface advancement with distraction osteogenesis for craniofacial reconstruction of a pycnodysostosis patient. A subcranial Le Fort III with a previously described distraction osteogenesis protocol was sufficient to provide bone stability to the craniofacial skeleton for this particular patient. However, further studies with a larger cohort may analyze the average individual bone physiologic response to this operation. Because pycnodysostosis is a very rare disease, a multicenter study may be required.

REFERENCES