Le Fort III Bipartition Osteotomy to Treat a Rare Craniofacial Anomaly: Frontofacionasal Dysostosis

Gökhan Tunçbilek, MD, PhD,* Yasemin Alanay, MD,† and Aycan Kayıkçıoğlu, MD*

Abstract: Frontofacionasal dysplasia or dysostosis (Mendelian Inheritance in Man [MIM] 229400) is composed of cranial, ophthalmic, nasal, and lip and palate deformities. It is a rare and separate entity because it contains none of the extracranial defects seen in frontonasal dysplasia and related syndromes.

Four patients with frontofacionasal dysplasia were treated in Hacettepe University between 2000 and 2007. A new surgical approach, less invasive and risky than an intracranial procedure and more effective and quicker than an orthodontic molding/distraction therapy, was developed during the treatment course of this group of patients. Le Fort III bipartition osteotomy was performed to reduce the distance between maxillary segments in very wide cleft lip and palate repair.

No surgical complication was encountered, and all the patients healed well and were discharged from the hospital without a problem.

Craniofacial deformities of the patients with frontofacionasal dysplasia should be repaired to have a socially acceptable facial appearance. We suggest Le Fort III bipartition osteotomy in selected cases to reduce the distance between maxillary segments in patients with very wide clefts.

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Frontofacionasal dysplasia or dysostosis (Mendelian Inheritance in Man [MIM] 229400) is composed of cranial (encephalocele, cranium bifidum occultum), ophthalmic (anophthalmia, blepharophimosis, telecanthus, S-shaped palpebral fissures, eyelid anomalies), nasal (deformed nostrils, nasal hypoplasia, bifid nose), and lip and palate (clefting) deformities. It is a separate entity because it contains none of the extracranial defects seen in frontonasal dysplasia and related syndromes. In addition, patients had more and severe craniofacial abnormalities than are seen in frontonasal dysplasia.

We report 4 additional patients with frontofacionasal dysplasia presenting with bilateral cleft lip and palate, bilateral anophthalmia, nasal deformities, and cranial bone clefts/defects.

PATIENTS AND METHODS

Four patients with frontofacionasal dysplasia were treated in Hacettepe University between 2000 and 2007. Patients were evaluated by a pediatric geneticist also. Common phenotypical findings in all 4 were bilateral cleft lip and palate, bilateral anophthalmia, nasal deformities, and cranial bone clefts/defects (Fig. 1). Location of the facial and cranial clefts resembled a Tessier class 1–13 cleft. Consanguinity was present in 2 families where there was also history of previously affected sibs. Patient history and craniofacial and extracranial features of the patients are presented in Table 1.

Surgical Techniques

Surgeries performed on the group of patients are listed in Table 2. Patient 1 had a large frontal bone defect with a wide bilateral cleft lip and cleft palate. Bipartition osteotomy with paramedian bone resection was performed in this patient to facilitate the repair of the wide cleft lip. The cranial defect was repaired at the same session using solvent-dehydrated calvarial bone allografts. In patient 2, orthodontic molding therapy was planned to replace the premaxilla and to lessen the gap between maxillary segments before the repair of the cleft lip and palate. An introral distractor, in its maximally distracted position, was attached to 2 introral plates each connected to the maxillary segments using stainless steel surgical wires. Although effects of reverse distraction procedure seemed less than expected, cleft lip repair was performed without problem.

Experiences gathered in these surgeries directed us to search another solution to facilitate the repair of the very wide cleft lip and palate. New solution should be less invasive and risky than an intracranial procedure and more effective and quicker than an orthodontic molding/distraction therapy. Patient 4 had a very wide bilateral cleft lip and palate with a protruded and superiorly located small premaxillary segment. An original solution, Le Fort III bipartition osteotomy, was planned (Fig. 2). Osteotomies were performed via transconjunctival and upper buccal sulcus incisions. Midface segments were approached to each other easily, and this made the bilateral cleft lip repair possible at the same session using modified Millard technique (Fig. 3). The whole procedure was completed in 3 hours.

RESULTS

All of the patients operated on with different techniques healed well and were discharged from the hospital without a problem (Fig. 4). No surgical complication was encountered. The mother of the first patient had another pregnancy, and during follow-up,
ultrasonographic examination showed a fetus with the same deformity. Pregnancy was terminated. Patient 2 later died of a respiratory infection at home 3 months after discharge.

DISCUSSION

Gollop suggested that a disorder observed in a brother and sister with consanguineous parents represented a “new” autosomal recessive disorder, which he called frontofacionasal dysostosis, in 1981. They reported an additional case with consanguineous parents in 1984. The fourth case of this disorder was reported by White et al in 1999. Frontofacionasal dysplasia is thought to be a result of the failure of normal development of the nasal capsule at about 33 days of gestation, which allows the primitive forebrain to move forward into space normally filled by the developing olfactory system. This leads to a cascade of anomalies with subsequent malpositioning.

The differential diagnosis of frontofacionasal dysplasia includes other syndromes in which facionasal dysplasia can appear. There are several syndromes in the literature under a variety of names. Phenotypical findings in the group of patients presented here are consistent with the findings listed for frontofacionasal dysplasia. None of our patients had extracranial defects as seen in frontonasal dysplasia (Online Mendelian Inheritance in Man [OMIM] 136760), craniofrontonasal syndrome (OMIM 304110), and acrofrontofacionasal dysostosis syndrome (OMIM 201180). The facial dysmorphic features are remarkably more severe than the other aforementioned entities. The history of affected sibs in families where consanguinity is present supports previous observations suggesting autosomal recessive inheritance as the mode of transmission in frontofacionasal dysostoses.

TABLE 1. Demographic and Physical Findings of the Patients

<table>
<thead>
<tr>
<th>Patient</th>
<th>Age, mo</th>
<th>Sex</th>
<th>Consanguinity</th>
<th>Family History</th>
<th>Craniofacial Findings</th>
<th>Extracranial Findings</th>
<th>Peripheral Blood Karyotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>11</td>
<td>M</td>
<td>+</td>
<td>+</td>
<td>Bilateral cleft lip and palate, bilateral anophthalmia, hypertelorism, coloboma of eyelids, frontal bone defect, protruded premaxilla</td>
<td>None</td>
<td>NA</td>
</tr>
<tr>
<td>2</td>
<td>6</td>
<td>F</td>
<td>-</td>
<td>-</td>
<td>Bilateral cleft lip and palate, right microophthalmia, left corneal opacity, corpus callosum hypoplasia, hypertelorism</td>
<td>None</td>
<td>NA</td>
</tr>
<tr>
<td>3</td>
<td>15</td>
<td>M</td>
<td>-</td>
<td>-</td>
<td>Operated bilateral cleft lip, cleft palate, hypertelorism, bilateral severe microophthalmia, bilateral orbital preseptal cysts, frontal bone defect</td>
<td>None</td>
<td>NA</td>
</tr>
<tr>
<td>4</td>
<td>10</td>
<td>M</td>
<td>+</td>
<td>+</td>
<td>Bilateral cleft lip and palate, bilateral anophthalmia, hypertelorism, frontal bone defect, protruded premaxilla</td>
<td>None</td>
<td>46, XX</td>
</tr>
</tbody>
</table>

M indicates male; F, female; NA, not analyzed.

TABLE 2. Surgical Procedures Performed to the Patients

<table>
<thead>
<tr>
<th>Patient</th>
<th>Frontal Bone Defect Repair</th>
<th>Cleft Lip Repair</th>
<th>Cleft Palate Repair</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Bipartition osteotomy with paramedian bone resection</td>
<td>Modified Millard repair</td>
<td>V-Y pushback palatoplasty</td>
</tr>
<tr>
<td>2</td>
<td>—</td>
<td>Modified Millard repair after orthodontic molding therapy</td>
<td>—</td>
</tr>
<tr>
<td>3</td>
<td>—</td>
<td>Operated elsewhere</td>
<td>V-Y pushback palatoplasty</td>
</tr>
<tr>
<td>4</td>
<td>—</td>
<td>Modified Millard repair after Le Fort III bipartition osteotomy</td>
<td>V-Y pushback palatoplasty</td>
</tr>
</tbody>
</table>
Le Fort III advancement osteotomy has been a major advance in craniofacial reconstruction. Midface advancement with a Le Fort III procedure is an important step in the treatment of the craniofacial dysostoses. Although it was first reported by Gillies and Harrison in 1951, Tessier refined the technique to the point of practical clinical application. Although Le Fort III procedure is mainly used for anterior and inferior movement of the midface, we modified it to facilitate the closure of very wide cleft lip with protruded premaxillary segment in patient 4. Premaxillary segment was placed such a superior and anterior position that lateral lip and cheek flaps should advance much more distance than a regular bilateral cleft patient. Wide undermining of the lateral flaps were performed as routine part of the cleft lip repair, but it seemed necessary to move zygomaticomaxillary segments with the soft tissues toward midline. This maneuver reduced the distance between 2 maxillary processes remarkably, and cleft lip closure was performed without difficulty. This maneuver facilitated also the cleft palate closure, performed 2 months later.

Another solution to facilitate the closure of the wide bilateral cleft lip with protruded premaxilla might be a premaxillary setback procedure. It has been shown that premaxillectomy with resection of the premaxillary-vomerine suture, in complete bilateral cleft lip and palate patients, causes flattening or concavity of the midface, loss of support for the upper lip, and failure of forward growth of nose. Premaxillary setback procedures performed early in the childhood resulted in midface retrusion and concave profile in adulthood. Projection of the nose in our patient was limited even before the cleft lip repair, so instead of a premaxillary setback procedure that might adversely affect the future growth of the midface, we mobilized the lateral maxillary segments to facilitate the cleft lip and palate repair. In conclusion, frontofacionasal dysplasia is a severe craniofacial malformation without extracranial deformities. Craniofacial deformities of these patients should be repaired to have a socially acceptable facial appearance. We suggest hemi–Le Fort III osteotomies in selected cases to reduce the distance between maxillary segments in very wide cleft patients.

**REFERENCES**