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Distraction osteogenesis therapy in patients affected by Goldenhar syndrome: a case series

ABSTRACT

Background Hemifacial microsomia (HM) is a syndrome characterized by the presence of structural alterations of the skeletal, nervous, vascular, and muscular structures derived from the first and second branchial arch. Goldenhar syndrome (Gs) consistisof the triad of craniofacial microsomia, ocular dermoid cysts, and spinal anomalies. When the patient has hypoplasia of the mandible, orthognatic surgery or distraction osteogenesis (DO) can be used to correct the asymmetry. Mandibular DO has been applied for many years, but long-term reports showed controversial results. The aim of this paper is to describe three cases of patients affected by Gs in which DO was performed to correct the mandibular asymmetry.

Case series The cases reported show an increasing degree of dismorphism which required a increasing complexity of the surgical approach: a single mandibular DO in the first patient, and a mandibular DO associated with a Le Fort I osteotomy in the second one, a double mandibular DO associated with Le Fort I and surgical disjunction of the middle palatal suture in the third case.

Discussion The effects of DO involve not only the skeletal segment but also all the surrounding soft tissues. DO leads to rapid and remarkable improvement in facial symmetry due to emimandible hypoplasia. When correct spatial repositioning of the maxilla cann ot be expected, mandibular DO can be carried out by associating a Le Fort I osteotomy. In this way DO minimize the need for major osteotomies and allows an earlier treatment in selected cases.

KEY WORDS Bone; Distraction osteogenesis; Goldenhar syndrome; Orthognathic surgery

INTRODUCTION

Hemifacial microsomia (HM) is a syndrome characterized by the presence of structural alterations of the skeletal, nervous, vascular, and muscular structures derived from the first and second branchial arch. The deformity may present in several different ways, ranging from severe cases of craniofacial dysmorphism to the more modest forms in which only the soft tissues are involved (1).

Goldenhar syndrome (Gs) is a rare condition that involves structures arising from the first and second branchial arches. It was first described in 1952 by Goldenhar and was later included in a broader classification called "Oculo-auriculo-vertebral spectrum" (2). Its incidence rate ranges from approximately 1:3000 to 1:5000 live births, it is more common in males, with a male-female ratio of 3:2. The patients right side of the face, body or both is generally more commonly and severely affected than the left (3). Gs consisting of the triad of craniofacial microsomia, ocular dermoid cysts, and spinal anomalies (4). It may also present heart diseases (5-58% of the patients)(5), hypoplasia of the zygomatic, mandibular and maxillary bones, muscle hypoplasia, anatomical and morphological abnormalities of the tongue, vertebral anomalies, cleft palate, disturbance of the central nervous system and other visceral anomalies (6, 7). Craniofacial anomalies, including mandibular, zygomatic and/or maxillary hypoplasia are found in 50% of patients with Gs (8).

When the patient has hypoplasia of the mandible, orthognathic surgery or distraction osteogenesis (DO) can be used to correct the asymmetry (9). In planning the correct therapeutic approach, it is necessary to bear in mind the degree of the deformity and the age of the patient. DO is used mainly in patients of developmental age and leads to the correction of the primary deficit as well as the morphologic recovery of many secondary

alterations (10, 11). To obtain good results, it is extremely important during treatment planning to define the aesthetic, structural, and functional aims that one intends to achieve with bone distraction. The effects of the distraction involve not only the skeletal segment, which is stimulated to lengthen gradually by itself, but also all the surrounding soft tissues such as muscles, skin, nerves, and vessels. Expansion of the soft tissues of the face leads to a rapid descent of the labial commissure, horizontalization of the chin, increase in volume of the distance between the labial commissure and the external orbital margin, and an increase in volume of the soft tissue of the cheek; all together, these changes result in a remarkable improvement in facial symmetry (12-14). When correct spatial repositioning of the maxillary cannot be achieved, mandibular DO can be carried out by associating a Le Fort I osteotomy (1). Mandibular DO has been applied for many years, but long-term reports present controversial results (3, 15). The aim of this paper is to describe three cases of patients affected by Gs in which DO was used to treat the mandibular asymmetry.

CASE SERIES

All patients were referred to the Department of Maxillofacial Surgery, Galeazzi Hospital, Milan (Italy).

Case 1

MD, a 7-years old boy, was seen in June 2001 since he had a right HM.

The aesthetic assessment revealed asymmetry of the face with hypoplasia of the lower third, including a hypoplastic mandible and an inferiorly and anteriorly displaced right ear lobule. Normal growth of the unaffected side accentuated the deformity and shifted the mandibular midpoint toward the affected side (Fig. 1). The radiological examination revealed the degree of mandibular asymmetry (Fig. 2). The facial features were consistent with a diagnosis of Gs.

The patient underwent DO by means of intraoral access. Under general anesthesia, an incision was made in the lateromarginal mucosa of the right mandibular angle. Subperiosteal detachment was performed with exposure of the gonial angle and of the area adjacent to the ascending portion of the right branch. Osteotomy of the mandibular branch was carried out above the last molar. The osteomized bone segments were mobilized and a 15 mm intraoral bone distractor was positioned. It was activated once a day with a 0.75 mm distraction for 9 days. Then the distraction device was maintained in place for 3 months in order to stabilize the area (Fig. 3, 4). Finally it was removed.

The patient gained a symmetrical aspect (Fig. 5). In October 2007 an Herbst device was used for 7 months to stimulate mandibular bilateral growth.

Fig. 1 *Frontal view of the patient at admission.*

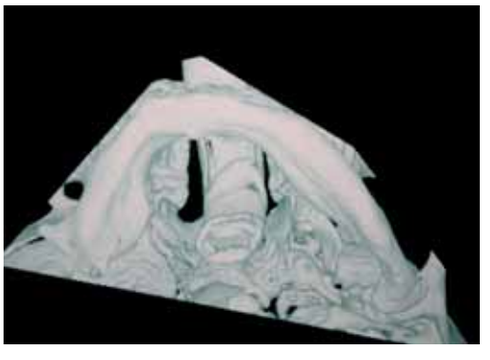

Fig. 2 *Down-up view of a CT 3-dimensional reconstruction highlighting the asymmetry of the mandibular arch.*

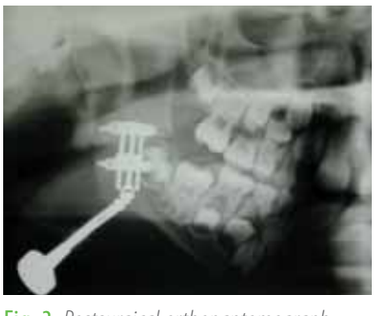

Fig. 3 *Postsurgical orthopantomograph showing the distraction device screwed across the osteotomic line.*

Fig. 4 *Orthopantomograph at 3 months: the distractor has reached the maximum expansion and the screw was cut (below) to give more comfort to the patient.*

Fig. 5 *Front view of the patient at the end of the follow-up (the right ear is not reconstructed yet).*

Case 2

SC, a 22-years old female, was evaluated in April 2002 since she had a left HM including asymmetry of the face due to hypoplastic mandible, deviation of the chin of the same side (Fig. 6) and inclination of the occlusal plane (Fig. 7). To better quantify the facial skeleton asymmetry, the patient underwent radiographic examination: orthopantomograph, telecranium radiograph in two projections and CT of the facial complex.

The patient underwent a DO by means of an intraoral access (Fig. 8), as previously described. After 4 months from the first operation, the distraction device was removed and in the same time a Le Fort I was performed (Fig. 9).

The patient gained a symmetrical aspect that was stabilized with a 8 months orthodontic treatment (Fig. 10, 11).

Fig. 6 *Front view of the patient at the first visit.*

Fig. 7 *Occlusion at the beginning of the treatment: the occlusal plane is deviated.*

Fig. 8 *Teleradiography performed at the end of the distraction period.*

Fig. 9 *The Le Fort I osteotomy.*

Fig. 10 *Front view of the patient at the end of the follow-up period: there is still an asymmetry when the patient smiles due to the soft tissues.*

Fig. 11 *The occlusion at the end of the treatment: see the correct position of the occlusal plane.*

Case 3

SI, an 18-years old female, was admitted in June 2005 since she had a left HM. Facial asymmetry was mild in rest position of the mimicking muscle (Fig. 12) but it remarkably increased when she smiled.
An endo-oral DO was performed by using two mandibular distraction devices, one for the ramus and one for the body (Fig. 13). In the same operation a Le Fort I osteotomy was also performed (Fig. 14) as well a midline palatal osteotomy with a subsequent position of maxillary distractor (Fig. 13). The patient gained a symmetrical aspect that was stabilized with a 12 months orthodontic treatment (Fig. 15).

DISCUSSION

Restoration of facial symmetry in HM, especially when it is associated with a soft-tissue deficiency, continues to be a difficult and challenging procedure for craniofacial surgeons (15).
Today, the DO, first described for orthopedic surgery by Ilizarov in the 60's, has minimized the need for major osteotomies and has allowed for earlier treatment in selected cases (15). DO has introduced a new concept for treating bone deficiencies since bone is formed in the area of the osteotomy by a tension-stress effect created by the distraction device (16). The distraction between the two bone segments is made with a controlled rate of increasing distance, usually of 1 mm per day for a maximum of about 10 mm in the facial region. Bone segment distraction leads to a formation of parallel columns of bone which extend from both edges to a central growth zone (17). The bone formed at 6 weeks has qualities of epiphyseal and intramembranous ossification (18). The peculiarity and uniqueness of this technique are that the bone generation is accompanied by a simultaneous expansion of the surrounding soft-tissue envelope, which contributes to the stability of the reconstruction and lessens the risk of relapse (19). The factors contributing to the success of this procedure are the thin layer of subcutaneous tissue involved, the minimum movement of the cutis, good vascularization of the soft tissues, and good healing that ensues (1).
All together the above mentioned concepts explain why DO is used in patients affected by facial malformation.
In Gs DO has been used since the early 90's (20).

Fig. 12 *Front view of the patient at admission.*

Fig. 13 *Orthopantomograph performed after surgery: there are two mandibular and one palatal distractors.*

Fig. 14 *The Le Fort I osteotomy: it was a classical osteotomy in the left side and a High Le Fort I in the right side.*

Fig. 15 *Front view of the patient at the end of the follow-up period.*

External (i.e. transcutaneous) devices were used at first, but they produced facial scars. Subsequently intraoral appliances, similar to those reported, were used and better results were obtained. Intraoral devices were used in the presented cases: they give satisfactory results avoiding the formation of facial scars.

The case series reported demonstrate that DO performed with intra-oral devices is an effective technique to restore facial symmetry and correct occlusion. DO has the advantage to distract not only the bone but also the surrounding soft tissues, avoiding the tension which is the major cause of relapse.

DO can also be performed in children, thus leading to use the growth potentiality of younger patient to correct their anomalies. Additional orthopedic and orthodontic treatment can be applied in association with DO and therefore DO performed in child reduces the need of additional major operations in adults.

In conclusion, DO is the treatment of choice in patients with facial malformations especially in asymmetric defects and during growth. Additional orthopedic and orthodontic treatments can be successfully applied to implement the DO results. Our data give additional strength to these guidelines.

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