Orthognathic surgery for management of Arthrogryposis Multiplex Congenita: Case report and review of the literature

Jennifer S Kargel BS, Vanessa M Dimas BS, Peter Chang MD DMD

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Arthrogryposis Multiplex Congenita is a condition characterized by multiple contractures of the joints. Involvement of the temporomandibular joint is a common complication that limits mandibular opening. A case of Arthrogryposis Multiplex Congenita with anterior open bite and limited maximal incisal opening is presented. Orthognathic surgery, consisting of segmental LeFort I osteotomy and bilateral sagittal split osteotomy, was performed successfully on this patient.

Key Words: *Arthrogryposis Multiplex Congenita; Orthognathic surgery; Temporomandibular joint*

The term Arthrogryposis Multiplex Congenita (AMC) refers to a congenital disorder first described by Otto in 1841 that is characterized by contracture of multiple joints, dimpling of the skin over the affected areas, cylindrically shaped extremities and an increase in fibrous tissue content of the muscles (1). The exact nature of the etiology of this disease is unknown, although it has been suggested that the likely cause is damage to the fetal anterior horn cells in utero. This leads to fetal paralysis and inadequate development and function of joints, often described as fetal muscle cicatrization. The incidence of AMC is low, occurring in only one per 5000 to 10,000 births, and it is usually not detected genetically. Patients often have severe contraction deformity of the shoulders, elbows, hips, knees and ankles. Surgical treatment and physical therapy aid in the reduction of these deformities, as well as improvement in joint function and alignment.

A review of the literature has shown that while both upper and lower extremities are usually involved, the temporomandibular joint (TMJ) is only affected in 25% of these patients (2). In AMC patients with maxillofacial involvement, manifestations may include micrognathia, limited jaw opening, high vaulted and/or cleft palate, and weakness of the muscles of mastication (3,4). Previously, it was thought that surgical treatment would not benefit patients with TMJ involvement due to the belief that hypomobility of this joint was secondary to muscular weakness and not a bony joint problem. In these patients, limited range of motion and opening of the jaw leads to dysfunctional speech, mastication and oral hygiene. To date, there have only been three reported cases of surgical treatment of TMJ manifestations of AMC in the surgical literature (2,3,5).

Une chirurgie orthognathique pour la prise en charge d'une arthrogrypose congénitale multiple : Rapport de cas et analyse bibliographique

Une arthrogrypose congénitale multiple est une affection caractérisée par de multiples contractions articulaires. L'atteinte de l'articulation temporomandibulaire est une complication courante qui limite l'ouverture mandibulaire. Un cas d'arthrogrypose congénitale multiple accompagnée d'une bécane antérieure et d'une ouverture incisive maximale limitée est présenté. Une chirurgie orthognathique, constituée d'une ostéotomie segmentaire de LeFort I et d'une ostéotomie sagittale bilatérale par clivage, a été exécutée avec succès chez ce patient.

Some methods used in these surgeries included bilateral coronoidectomies, TMJ arthroplasty and bilateral condylectomies. A case of AMC with anterior open bite and limited maximal incisal opening (MIO) is presented, and the successful orthognathic surgical treatment methods are described.

CASE PRESENTATION

A 16-year-old girl with a history of diffuse AMC presented to the clinic with the chief complaint of limited mouth opening and difficulty chewing hard foods, caused by an anterior open bite and limited MIO. At the time of presentation, orthodontic treatment had been ongoing for three years. Evaluation of the patient revealed malpositioned dentition both within and between each arch, as well as an abnormal skeletal relationship of the upper and lower jaws (Figure 1). The treatment plan included continuation of aligning of the dentition within each arch with orthodontics, followed by restoration of the maxillary-mandibular dental relationship with skeletal osteotomies and repositioning. At the age of 18 years, she was again evaluated, and it was determined that the dentition was in good alignment in the arch (Figure 2), and treatment to include correction of the skeletal deformities should begin. The plan was to widen the maxilla to correct the cross-bites and to posteriorly impact and advance the maxilla to close the anterior open bite. Additionally, the mandible would be set back with a bilateral sagittal split osteotomy (BSSO) and autorotate to fit into occlusion with the maxilla. Preoperative measurements were a 15 mm open bite and an MIO of 25 mm, with only 10 mm of functional mobility.

University of Texas Medical Branch, Galveston, Texas, USA

Correspondence and reprints: Dr Peter Chang, Cosmetic Surgical Institute, PO Box 1357, League City, Texas 77574, USA.

Telephone 409-772-1255, fax 409-772-1872, e-mail drpetechang@gmail.com

Figure 1) Presurgical and preorthodontic radiographs of patient. **Left** Lateral view; **Right** Panoramic view

Figure 2) Presurgical clinical views of patient. **A** Frontal view of mouth; **B** Frontal view; **C** Right lateral view

This patient underwent a midline split segmental LeFort I osteotomy with 8 mm of posterior impaction, and 8 mm of advancement on the right and 6 mm of advancement on the left. She also underwent a BSSO with 3 mm of setback, partial bilateral inferior turbinectomies and bilateral coronoidectomies. The MIO was increased from 25 mm to 31 mm. The operation proceeded smoothly and without complication. The postoperative course remained uneventful, and one year postoperatively, she remained pleased with the results of her surgery (Figures 3 and 4).

DISCUSSION

Most reported AMC cases in the scientific literature focus on the management of limb deformities involving the upper and lower extremities. A literature search has identified only three previously reported cases of surgical correction of AMC involving the maxillofacial area (2,3,5). Previous reports claimed that maxillofacial surgery would not benefit AMC patients, because it was believed that the restrictions associated with this type of joint hypomobility were the result of muscular deficits and not skeletal abnormalities (6,7).

Epstein and Wittenberg (2) reported the first case of AMC managed with maxillofacial surgery. They presented the case of a 21-year-old woman with AMC who had class I malocclusion, mild anterior open bite and a severely restricted interincisal opening of 12 mm. Surgical treatment included bilateral coronoidectomies, a left TMJ arthroplasty, high condylectomy and temporal eminence reduction, as well as insertion of a teflon-proplast implant into the joint cavity at

the lateral rim of the glenoid fossa. The resulting MIO increased to 23 mm. The authors concluded that surgical management, supplemented by postoperative physiotherapy and stretching exercises, offered significant improvement in TMJ functioning.

The second case of surgically managed AMC involving the maxillofacial region was reported by Hodgson et al (5). These authors described a case of limited mandibular opening in a 30-year-old male patient suffering from arthrogryposis. Radiographic studies showed a normal right TMJ and a grossly deformed left TMJ lacking any joint space. Previously, this patient had received a blow to the mandible, causing a fracture that required open reduction and internal fixation. However, no trauma to either TMJ was reported at that time. Using a preauricular approach, a left condylectomy was performed. Within one week postoperatively, the patient's MIO had increased from 22 mm to 30 mm. They concluded that, although this patient suffered from AMC of the extremities, the deformity of the left TMJ was not typical of those reported in the previous literature describing AMC manifestations in the TMJ and may have been the result of an alternative cause.

Finally, a third case describing surgical treatment of AMC in the maxillofacial region was described by Thomas et al (3) in 2001. These authors suggested that many of the prior reports warning against surgical management of hypomobility of the TMJ, and stating that the defect is related to muscular weakness instead of a bony defect, had focused on younger patients in whom skeletal changes may not yet have been evident (4).

Figure 3) Postsurgical radiographs of patient taken one year postoperatively. **Left** Lateral view; **Right** Panoramic view

Figure 4) Postsurgical clinical views of patient. **A** Frontal view of mouth, taken at two months postoperatively; **B** Frontal view, taken at one year postoperatively; **C** Right lateral view, taken at one year postoperatively

In this case, a 28-year-old male patient suffering from AMC and limited mouth opening underwent bilateral intraoral coronoidectomies, meniscectomies, capsular release and lateral pterygoid myotomies. Postoperatively, the MIO increased from 9 mm to 18 mm. They suggested that a surgical approach, in addition to stringent postoperative mouth-stretching exercises, offers significant functional improvement in patients with AMC who experience limited mouth opening secondary to disease involvement of the TMJ.

Our case is unique in that it is the first reported use of orthognathic surgery in the treatment of maxillofacial manifestations of AMC. The combined surgical procedures of bilateral coronoidectomies, segmental LeFort I osteotomy and BSSO successfully gave our patient an effective functional MIO of 31 mm. By closing the anterior open bite and releasing the attachments at both coronoids, this patient was given more mobility of her jaw than any single procedure could have given her. Her speech and oral hygiene have also improved due to the correction of her open bite.

Distraction osteogenesis was initially considered for this case, but after a thorough workup with predictive cephalometric tracings and model surgery, our goal of closing the open bite and improving her MIO would not have been achieved expeditiously. Ideally, a rapid palatal expander may have helped in the correction of her narrow maxilla while she was receiving active orthodontic treatment, but due to financial restrictions, that option was not possible.

CONCLUSION

The use of orthognathic surgery for management of facial deformities in arthrogryposis has never been reported in the surgical literature. Based on our experience, this approach is a valuable tool that offers considerable functional improvement of mobility, mastication, speech and oral hygiene. This surgical procedure, along with postoperative physical therapy to maintain the MIO, should be discussed with the patient as a potential option for management of AMC.

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