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Arthrogryposis Multiplex Congenita in Association With Bilateral Temporomandibular Joint Hypomobility: Report of a Case and Review of Literature

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Arthrogryposis is a physical sign observed in many specific medical conditions. The Greek language translates arthrogryposis as "curved joint." Arthrogryposis multiplex congenita (AMC) is currently classified under a heterogeneous group of disorders featuring multiple congenital joint contractures. AMC can be classified into 3 main groups: disorders with mainly limb involvement, disorders with limb involvement together with involvement of other body parts, and disorders with limb involvement and central nervous system dysfunction. AMC is seen in approximately 1 in 3,000 to 10,000 live births without any gender predilection.[1-4] Family history and pregnancy history are considered to be plausible contributors but not definitive.[1,5,6] Intellectual underdevelopment tends to vary from patient to patient. Reports of mental underdevelopment range from nonexistent to an incidence as high as 10%.[1,5]

The principal causes of AMC disorder can be of fetal origin or can result from compromising maternal issues. Fetal neurogenic, muscular, or connective tissue abnormalities, which contribute to fetal akinesia (decrease in fetal movement) during gestation, are plausible causes. Fetal akinesia is generally accepted as the pathologic cause of AMC. Fetal akinesia results in extra connective tissue around the joint, causing fixation and further joint contracture.[1,3,5,7,8] Maternal illness, drug use, and trauma during gestation are also credible causes. Interestingly, joint development during early embryogenesis is almost always normal in AMC patients.[1,3,5,7,8]

There are numerous other syndromes and disorders that show multiple congenital joint contractures. Review of associated syndromes is not the focus of this report. However, syndromes having a possible association with maxillofacial manifestations include Marfan, Pierre-Robin, Down, Turner, Crouzon, Möbius, and mandibulofacial dysostosis.[1] Oral and maxillofacial manifestations include micrognathia, microstomia, hypomobility of the mandible, high-vaulted palate, cleft palate, and weak masticatory muscles.[1,8-10] In a previous study by Steinberg et al,[10] the 3 most common maxillofacial symptoms in AMC were reported to be mandibular hypoplasia, limited mandibular opening, and high-arched palate. Mandibular hypoplasia and limited mandibular opening have been well documented as being commonly associated with AMC.[1,3,7,9,10] A list of other abnormalities with or without the possibility of maxillofacial manifestations is listed in Table 1.

Table 1. AMC ASSOCIATIONS

Marfan syndrome
Bony fusion
Contractural arachnodactyly
Multiple pterygium syndrome
Freeman-Sheldon syndrome
Osteochondrodysplasia
Mandibulofacial dysostosis
Pierre-Robin syndrome
Cerebro-oculofacioskeletal syndrome
Chromosome abnormalities (ie, trisomies)

NOTE. The syndromes and disorders listed are not comprehensive. Individualized diagnosis and treatment planning are of the utmost importance. Data are modified from Epstein and Wittenberg[1] and Lloyd-Roberts and Lettin.[14]

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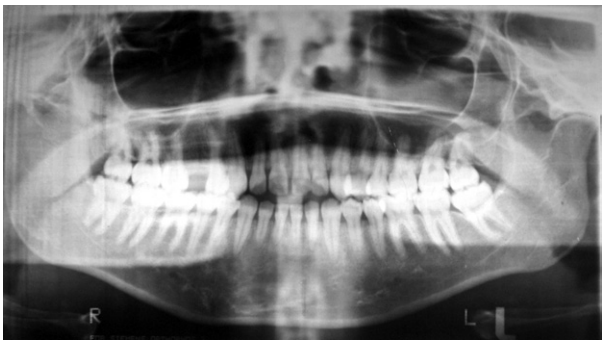

FIGURE 1. Panoramic radiograph at initial examination.

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Report of a Case

In 2003, a 23-year-old African-American woman presented to the outpatient office of the Division of Oral and Maxillofacial Surgery, Drexel College of Medicine, Philadelphia, PA, with the chief complaint of "cannot open mouth." The patient was diagnosed with AMC as a child and was otherwise healthy with a noncontributory family and social history. Her surgical history included bilateral hand and wrist joint surgeries, bilateral ankle surgeries, and bilateral hip joint surgeries.

The patient was surprisingly self-sufficient and currently enrolled in undergraduate studies. On physical examination, her maximal incisal opening (MIO) was no greater than 15 mm. There was an absence of lateral and protrusive movements of the lower jaw. A panoramic radiograph showed bilateral hypertrophic condylar head and absence of joint anatomy (Fig 1).

At that point, the patient was scheduled to undergo a head computed tomography (CT) scan and temporomandibular joint (TMJ) magnetic resonance imaging (MRI) for further assessment (Fig 2). The CT scan showed bilateral flattening and loss of cortex of the condylar heads. In addition, narrowing of bilateral TMJ space with saucerization of the glenoid fossa was reported. MRI showed degenerative changes in both TMJs with more severity on the right. Both TMJs showed little or no anterior translation upon mouth opening. A long and detailed discussion regarding the possibility of bilateral coronoidectomy and bilateral arthroplasty with meniscus disc replacement or reattachment was discussed. At that time, the patient consented to bilateral coronoidectomy with the possibility of bilateral arthroplasty. An emphasis was also placed on aggressive physical therapy (tongue blade and manual manipulation by the patient) and return for re-evaluation.

At the 1-month follow-up office visit before surgery, the patient's MIO was 25 mm versus the previous measurement of 15 mm. It was then decided that she should continue aggressive physical therapy and return for re-evaluation 1 month later. During the 2-month office visit, her MIO was measured to be 26 to 27 mm (approximately 14 tongue blades). Because of her continuing clinical improvement, we recommended continuing aggressive tongue blade therapy and considered bilateral TMJ debridement instead of coronoidectomy. Unfortunately, an emergent odontogenic infection developed in the patient. With existing limited access, the general dentist was unable to address her dental issues. The patient was scheduled for bilateral coronoidectomy and Brisement force at Hahnemann University Hospital (Philadelphia, PA).

The operation was performed without any complications. Intraoperatively, an MIO of 25 to 30 mm was achieved with Brisement force. The patient was discharged and instructed to return for follow-up on a routine basis. At 1 week after bilateral coronoidectomy, the findings were positive; the patient was doing well, with an MIO of 10 to 15 mm without manual assistance and 15 to 20 mm with manual assistance. Aggressive physical therapy was again emphasized. At 2 weeks after coronoidectomy, the MIO remained at 15 to 20 mm. We instructed the patient to continue physical therapy and return for follow-up 2 to 4 weeks thereafter. Examination at 3 weeks after coronoidectomy showed no additional MIO improvement with positive patient compliance regarding physical therapy. At that point, the possibility of bilateral TMJ surgery was discussed.

The patient returned to the clinic 1 month later with a growing interest in bilateral TMJ surgery with reconstruction. A 3-dimensional CT scan reconstruction was obtained to better visualize the existing TMJ anatomy (Fig 3). It was decided, with the consent of the patient, to perform bilateral open joint arthroplasty with possible condylectomy and placement of temporalis fascia. The patient was scheduled to undergo surgery in June 2005.

At 1 week after bilateral arthroplasty with temporalis fascia placement, we recorded an MIO of 28 mm, and the

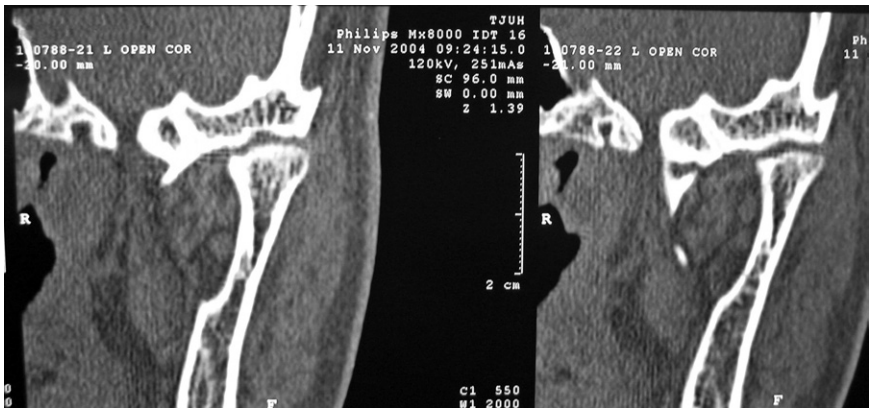

FIGURE 2. CT scan.

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patient claimed to have been continuously using the TheraBite system (Atos Medical, Hörby, Sweden) 3 times a day. At 2 weeks she showed no improvement in MIO with aggressive physical therapy. The MIO increased by 10 mm to 35 mm during the patient's third week of follow-up. At the fifth week of follow-up, an MIO of approximately 30 to 35 mm was recorded with increased edema of the right TMJ region without pain. Unfortunately, the patient did not return to the office until 1 month after the last visit, and her MIO decreased to 20 mm. At that point, we began to doubt her commitment and her claim of performing TheraBite therapy 3 times a day. Physical therapy was again emphasized, and the patient was asked to return 1 month later.

Upon the patient's return in March 2006, she had completely scarred down and could no longer open her mouth. At that time, she finally admitted to discontinuation of physical therapy and agreed to the possibility of requiring further surgery. She was again noncompliant in her follow-up sessions, and she returned to our office in November 2006 completely locked and unable to open her mouth at all (Fig 4). It should be noted that the patient's physical limitation could have contributed to her noncompliance.

Given the history of this patient, it was decided, with consent, to re-establish her TMJ space by performing bilateral gap arthrotomy and distraction with a bilateral craniomandibular fixator (Matthews Device; KLS Martin, Jacksonville, FL). The craniomandibular fixator was stabilized superiorly on the temporal bone and inferiorly on the ramus (Fig 5). This external device will not only re-establish a patient's TMJ space but also maintain joint space and allow for a possible increase in joint space as the device is activated. In addition, the hinge apparatus should promote greater joint mobility. Our final preparation for surgery included obtaining stereolithic models (Fig 6).

Intraoperatively, bilateral scar tissue, osseous union, and absence of TMJ space were evident. An MIO of 20 mm was achieved with Brisement force after bilateral gap arthrotomy (re-established joint space of 2-4 mm) (Fig 7). Postoperatively, the distraction device was activated bilaterally 1 mm/d by turning it in a clockwise manner.

Upon discharge, the patient was instructed to self-activate the distraction device 1 mm/d at home. At 1 week, the patient was doing well and had an MIO of 15 to 20 mm (Figs 8, 9). During her 2-week follow-up, she was able to maintain the operatively accomplished MIO of 20 mm. The patient was undergoing aggressive physical therapy and was on schedule for the removal of the bilateral TMJ distraction

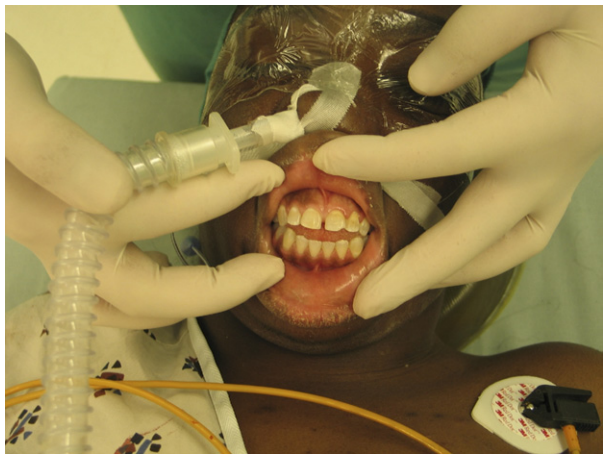

FIGURE 4. Presurgical presentation in November 2006.

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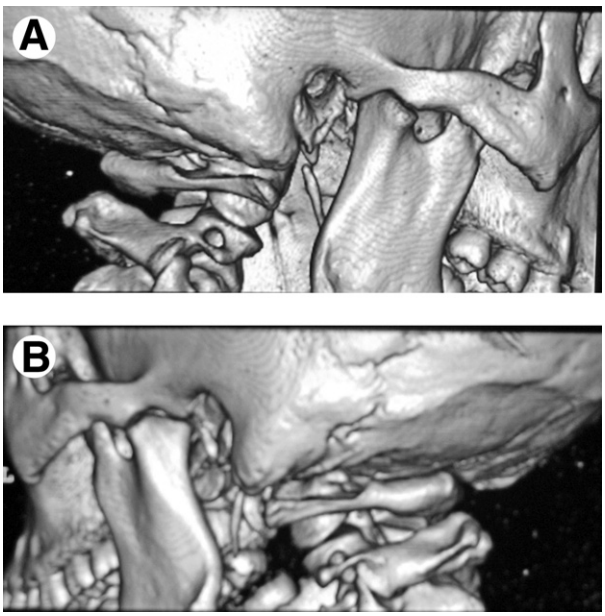

FIGURE 3. MRI scans of right TMJ (*A*) and left TMJ (*B*).

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FIGURE 5. Craniomandibular fixator (Matthews Device).

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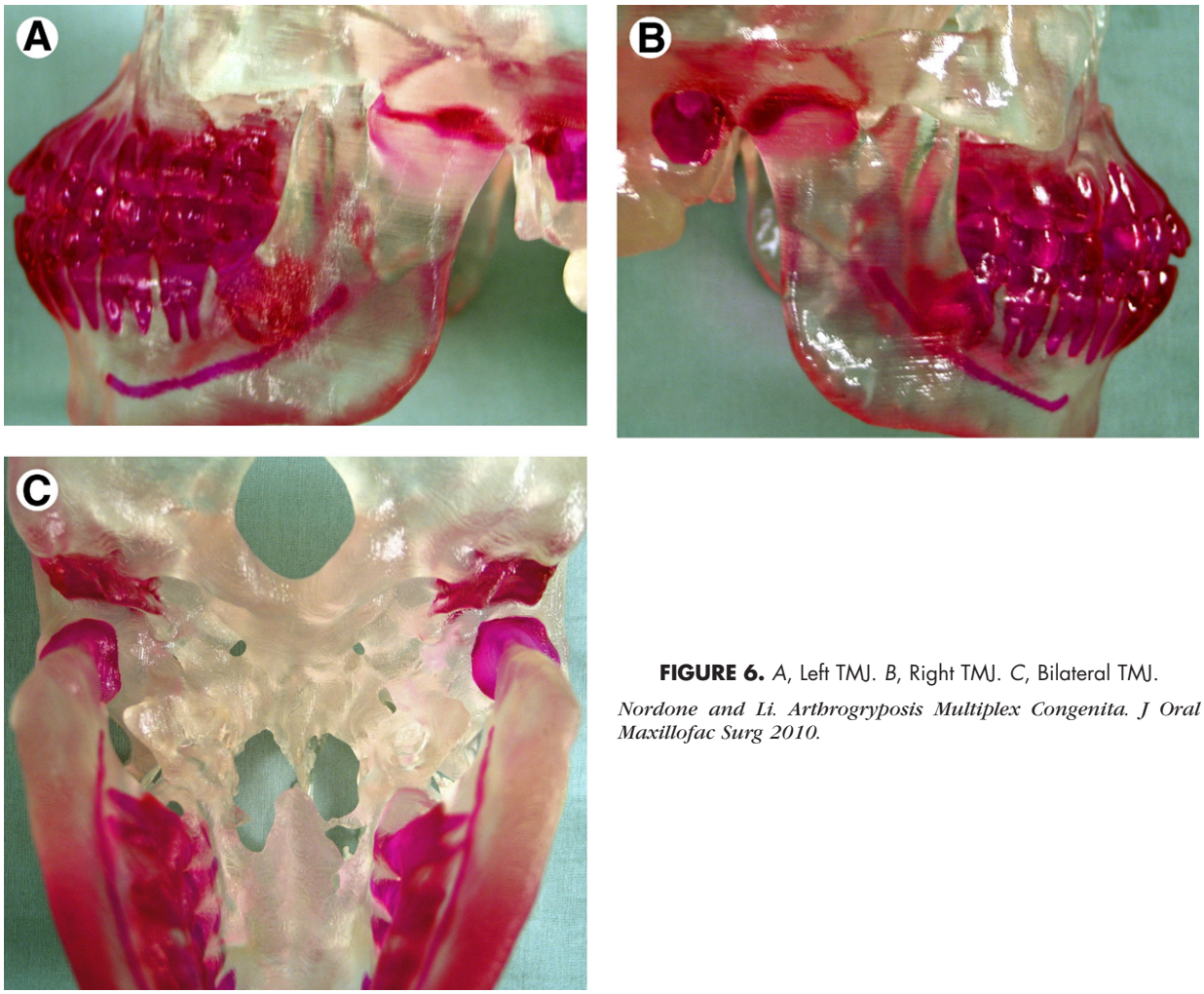

FIGURE 6. *A*, Left TMJ. *B*, Right TMJ. *C*, Bilateral TMJ.

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device. In total, the distraction device had been in place for 3 months and had significantly improved her masticatory functions (MIO of 25 mm). Figures 10 and 11 are the clinical pictures of the patient 2 weeks before the removal of the distraction device.

Discussion

In the AMC patient, physical examinations tend to show a wide array of findings. There are, however, several common characteristics to keep in mind. The involved extremities are cylindrical in shape with thin subcutaneous tissue and without skin crease. Deformities are noted to be symmetric and increase in severity distally; that is, the hands and feet are the most affected. Joint rigidity and dislocation may be present with atrophic or absent associated muscle groups.[4,9,11] Of special interest, the TMJ is frequently found to have limited range of motion.[10,12,13] Facial deformities may include the following: asymmetry, flat nasal bridge, hemangioma, micrognathia, trismus, and palatal deformities. Other physical deformities include scoliosis, genital deformities, hernia, pulmonary hypoplasia, hypertelorism, cleft palate, depressed nasal tip, high nasal bridge, functional short gut with feeding problems, and short umbilical cord.

Pathologically, there are 2 subcategories involved: neurogenic types and myopathic types of AMC. The most common neurogenic abnormalities in arthrogryposis are the nonspecific muscle fiber types.[5] As a result of the dysgenesis of the motor nuclei of the brain stem and spinal cord fasiculi, muscle fibers are replaced by small muscle fibers and adipose tissue. Examples include patients with Pierre-Robin syndrome and Möbius syndrome.[13] Dysgenesis of the central nervous system is the second most common neurogenic abnormality in arthrogryposis (23%), with disorganization of and a decrease in the number of neurons of the cortex and motor nuclei of the brainstem and spinal cord. Examples include trisomy 18, partial deletion of the long arm of chromosome 18, and Zellweger syndrome.[1,10]

In myopathic types of AMC, central core disease occurs, where the central portion of each muscle

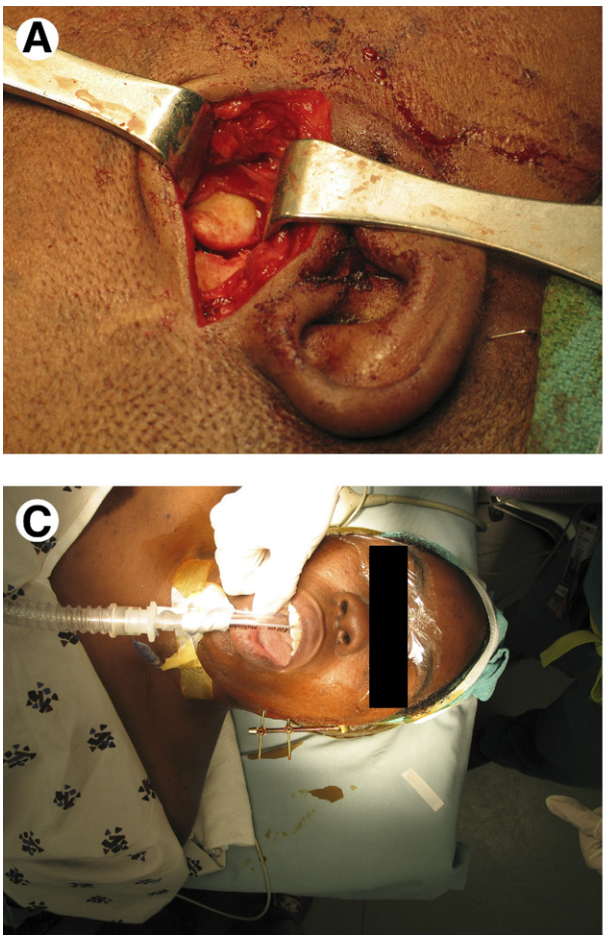

FIGURE 7. *A*, Right gap arthrotomy. *B*, Left gap arthrotomy. *C*, Immediate postoperative opening.

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fiber contains a zone in which oxidative enzyme activity is absent.[5] Congenital muscular dystrophy is indicated by muscle fibers that show a rounded configuration with variation in diameter. Perimysial and endomysial connective tissues are increased markedly.

Management

There is no definitively proven successful approach to manage AMC. Alignment and establishment of stability for ambulation and function for self-care are principles from orthopedic surgery

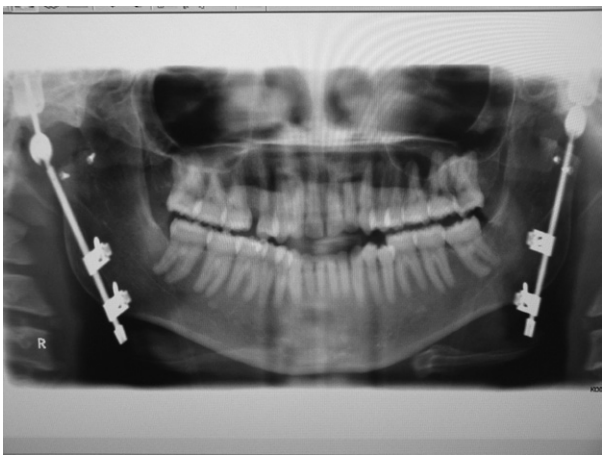

FIGURE 8. Panoramic radiograph at 1 week's follow-up.

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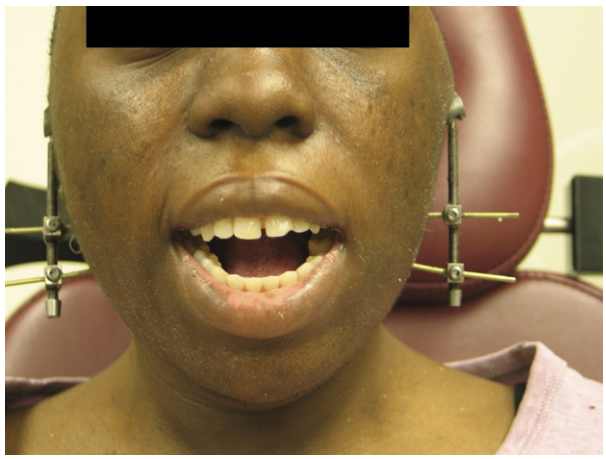

FIGURE 9. MIO at 1 week's follow-up.

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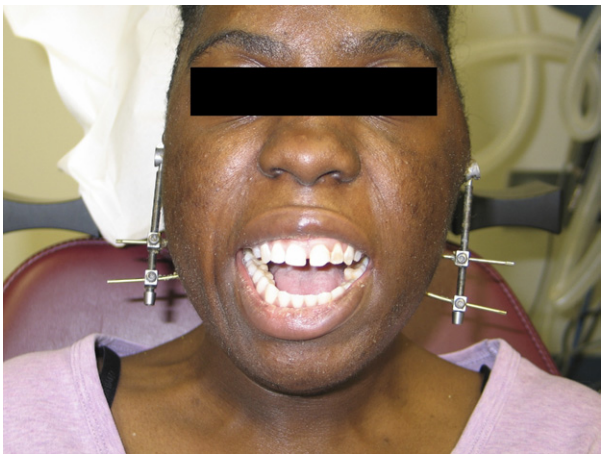

FIGURE 10. MIO at 10 weeks' follow-up with intensive physiotherapy, front view.

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that can be directly applied to the maxillofacial region. Williams[11] described 4 principles of treatment for most arthrogryposis joints: tendon transfer from a normal muscle to a hypoplastic muscle, surgical release of ligaments and muscles, prolonged immobilization, and intensive physiotherapy. Lloyd-Roberts and Lettin[14] suggested that capsulotomy followed by prolonged splinting could correct deformities of the long bones. Repeated surgeries may be necessary, and prolonged physiotherapy may become a necessity. Early manipulation soon after birth improves passive and active range of motion. Early vigorous physical therapy to stretch contractures is vital in improving joint motion and avoiding muscle atrophy.[1-15]

Treatment of the TMJ with arthrogryposis remains controversial and requires prudent surgical assessment. Hageman and Willemse[3] found that 25% of the patients diagnosed with AMC had TMJ involvement. A contradictory article reported that TMJ involvement was rare.[11] It is, however, generally accepted to expect a certain degree of TMJ involvement in AMC patients.

Previous reports have supported bilateral coronoidectomies to increase condylar translation and relief of interference and tension from hypoplastic or atrophic temporalis muscles.[2,7,8,10] Epstein and Wittenberg[1] reported treatment of limited mandibular opening with bilateral coronoidectomy, which failed, and subsequent treatment with TMJ arthroplasty with release of adhesions, a high condylotomy, and placement of a Proplast-Teflon disc implant, which improved mandibular function. Steinberg et al[10] resorted to rigorous continuous and long-term physiotherapy. The difficulty in establishing a treatment protocol for AMC patients with TMJ involvement remains, because of the limited number of patients with significantly limited range of motion.[2,10,12,13]

Of special interest, Gabbay et al[15] conducted a comparison study using transport distraction osteogenesis and Matthews Device arthroplasty to compare the long-term efficacy of the 2 procedures in pediatric patients diagnosed with TMJ bony ankylosis with micrognathia. Their concluding data showed that both procedures were initially successful surgically but patients undergoing Matthews Device arthroplasty showed significantly less relapse.

We believe that it is prudent to assess each patient based on functional capacity and esthetic concerns. If the patient is functionally self-sufficient and in good mental health regarding his or her physical condition, surgery is not indicated; rather, aggressive physiotherapy should be encouraged. However, if the patient cannot maintain adequate nutrition and growth, it is necessary to provide other treatment options. We recommend taking a step-wise approach in treating the patient. Figure 12 details the proposed treatment algorithm.

The principal cause of AMC has been repeatedly suggested as fetal akinesia. The end result of increased joint contracture and hypomobile joints has also been associated with muscle atrophy and/or hypoplasia. In contrast to these hypotheses, Guimaraes and Nagahashi[5] showed that there are no morphologic abnormalities in the TMJs or mastication muscles but, instead, that there are elongated coronoid processes that mechanically hinder jaw movements.

After evaluation of our 23-year-old African-American female patient in 2003, CT scan showed bilateral flatten-

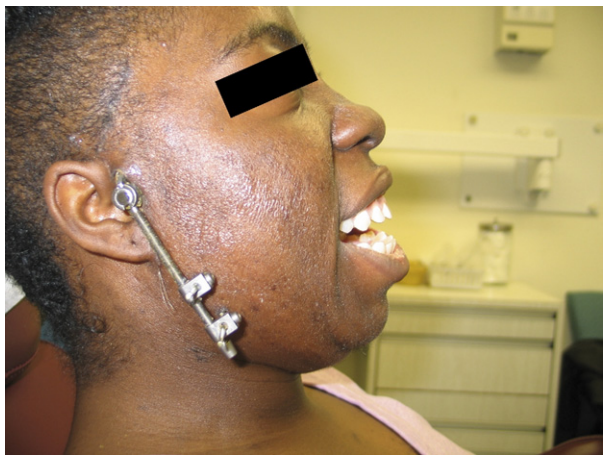

FIGURE 11. MIO at 10 weeks' follow-up with intensive physiotherapy, side view.

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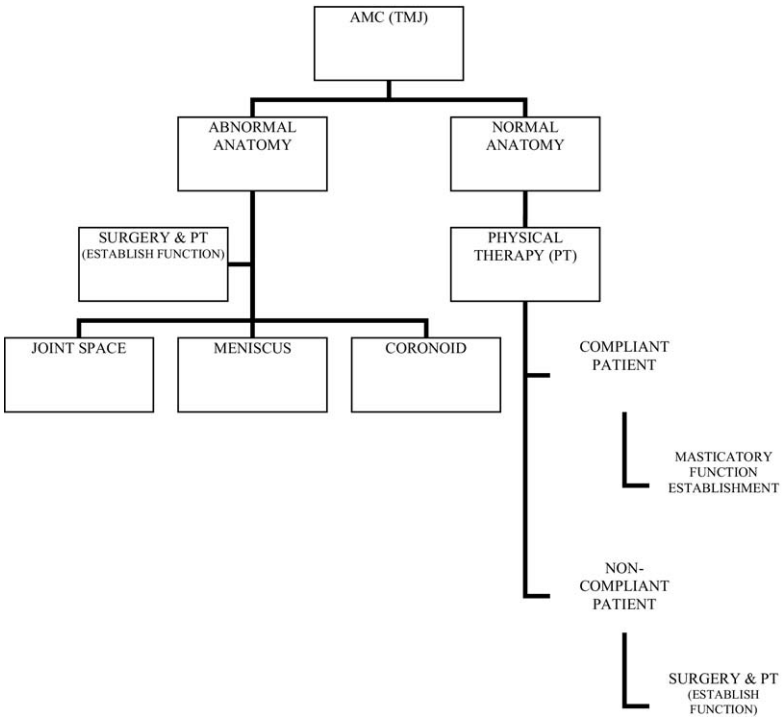

FIGURE 12. Proposed algorithm for management of AMC with TMJ involvement.

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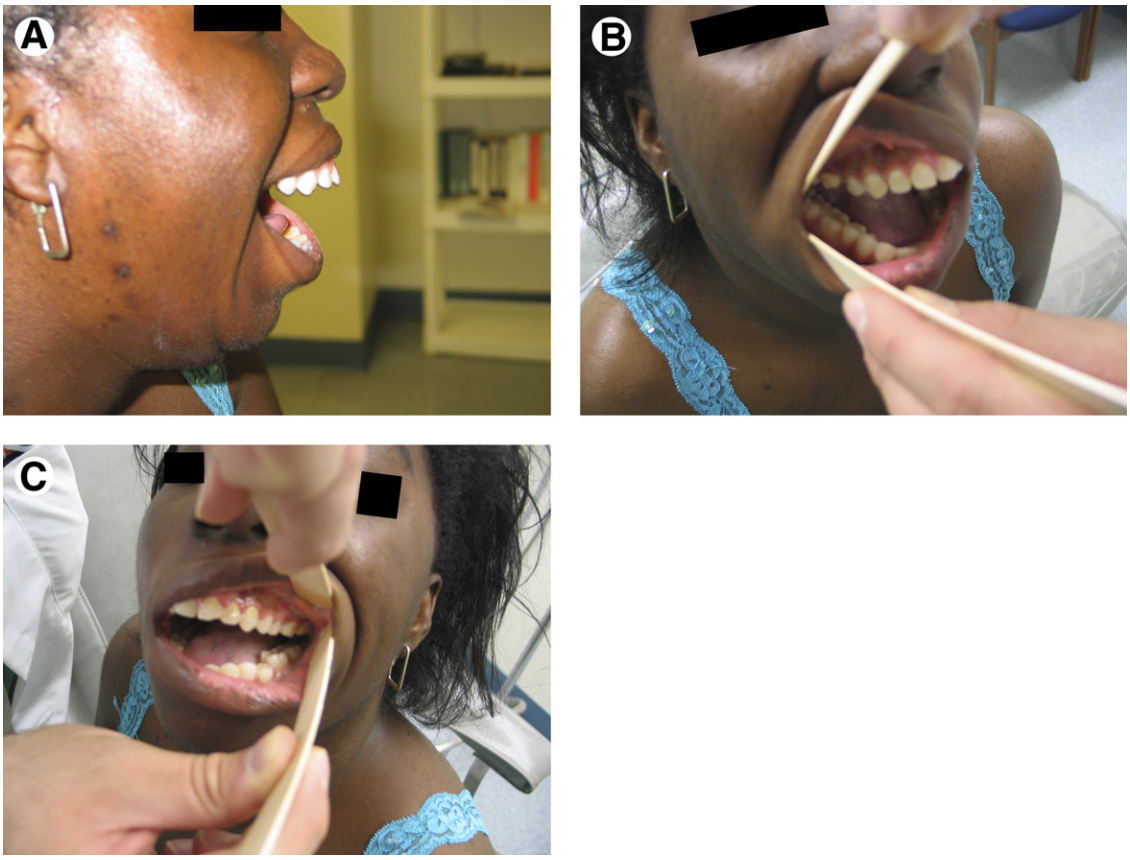

FIGURE 13. *A-C*, MIO at 2 months' follow-up.

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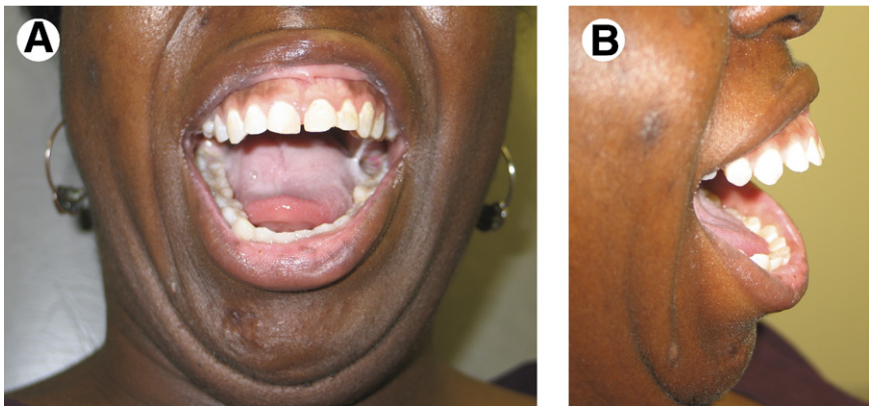

FIGURE 14. *A, B,* 1 year follow-up.

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ing and loss of cortex of the condylar heads and narrowing of bilateral TMJ space with saucerization of the glenoid fossa. MRI showed degenerative changes in both TMJs, with greater severity on the right. Both TMJs showed little or no anterior translation upon mouth opening. At 2 years after bilateral coronoidectomy, repeated CT scan and 3-dimensional reconstruction illustrated regrowth of both condyles into the infratemporal space. This reinforces the hypothesis of an osseous contribution to the restriction of mouth opening, but it cannot conclusively be the cause of our patient's hypomobile mandible because there is a coexistence of abnormal effects of fetal akinesia. Unfortunately, the patient was not seen at our institution since birth, and pathologic factors leading to this abnormality remain a mystery. One additional examination that may have been beneficial may have been electromyography after the initial office visit at the Division of Oral and Maxillofacial Surgery, Drexel College of Medicine. Perhaps electromyography could have elucidated 1 additional contributing factor to her TMJ condition.

Our patient is currently progressing well with reestablishment of MIO of 25 to 30 mm at her 2-month follow-up and MIO of 42 mm at her 1-year follow-up. The craniomandibular fixator was originally in place for a total of 4 months. Figure 13 shows the patient at 2 months' follow-up after removal of the craniomandibular fixator (Matthews Device). Figure 14 shows her at 1 year of follow-up after removal of the craniomandibular fixator (Matthews Device). She will continue aggressive physiotherapy because relapse is of major concern.

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