Maxillofacial Implications and Surgical Treatment of Arthrogryposis Multiplex Congenita: A Case Report and Literature Summary

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Abstract

A case of Arthrogryposis Multiplex Congenita which affected a patient's temporomandibular joint is described. Pre-operatively, the patient's interincisal opening was limited to nine millimeters. Elective surgery was performed, consisting of bilateral coronoidotomies, right and left meniscectomies, capsular release, and lateral pterygoid myotomies, and physical therapy was initiated postoperatively. Eighteen weeks following the surgery, the patient was able to open eighteen millimeters, and force open to twenty millimeters. The patient also noted significant improvement in speech and jaw function in the post-operative period.

Introduction

Arthrogryposis Multiplex Congenita, which has been more recently termed Multiple Congenital Contractures, may affect from 1 in 3,000 to 1 in 10,000 live births.[1,6,9,11] It has been shown to have a predilection for males, and in some forms possesses a genetic component.[9] It is rarely fatal, but can be crippling if surgical intervention does not occur at a young age. This article will seek to review some of the maxillofacial implications of this disorder, and also describes a case report. To our knowledge, this is only the third such case documented in which a

surgical approach has been utilized to treat the maxillofacial complications of a patient with this disorder.

AMC is really a collection of disorders which ultimately result from intrauterine fetal paralysis.[7] This paralysis can be caused by mechanisms native to the fetus itself or to its intrauterine environment. Mechanisms native to the fetus include both neurogenic factors and myopathic factors.[5,7,11] The former are characterized by a decrease in the size and number of fetal spinal anterior horn cells with denervation atrophy of associated musculature.[5,6,7,11] The latter may result from a variety of connective tissue disorders that prevent normal muscular development. Mechanisms associated with the intrauterine environment include IUVA, drug-induced fetal immobilization, increased or decreased amniotic fluid, the presence of twins, bi-cornate uterus, wrapping of the umbilical cord, poor placental circulation, ovarian cysts, and maternal viral infections,[1,6] specifically those associated with the Akabane virus during early pregnancy.[10,11] In all cases, fetal paralysis leads to the limitation or elimination of normal intrauterine fetal movement, which is essential in the development of normal joints and their associated skin creases.[10] Anatomic manifestations most commonly include multiple rigid joint deformities, usually affecting all four extremities.[7,11] Typically, when only two extremities are affected, it is usually the lower.[7] Extremities appear fusiform and cylindrical, with a tense or glossy appearance to the overlying skin, due to lacking skin creases.[5,6,7,11] Multiple joint dislocations are common, and muscle groups may be atrophied or altogether missing. As stated previously, the CNS may be affected, but intellectual development is usually normal in these patients.[5,7,10,11] Many other manifestations have been noted, but a discussion of these is beyond the scope of this article.

Maxillofacial abnormalities, though less common, have also been recently well-documented; the most common findings cited include decreased mandibular opening, mandibular hypoplasia, a vaulted palate, and deficient musculature of the oro-facial complex. Other findings which have been described are limited tongue protrusion, cleft palate, dysphagia, airway compromise, poor gag reflex, and hemangiomas of the upper face.[1,3,6,9]

Classically, surgery to correct temporomandibular abnormalities in these patients has not been recommended, because most cases demonstrated problems attributable to muscular insufficiencies.[2] Specifically, it has been suggested that hypomobility of the TMJ is not related to a bony defect, but rather to muscular weakness, and therefore surgery would not be indicated. Notably, however, one author points out that many studies have focused on younger patients in whom bony changes may initially not be evident.[9] Typically, these cases have been managed with physical therapy, which was shown to be successful until discontinued.[1,9] More recently, some cases have been treated surgically to correct limitations in opening. At least one case which succeeded in increasing the patient's maximum opening has been described in the literature by Epstein.[1] However, no previous cases have been described in which a patient who demonstrated the typical insufficiency of muscular strength on EMG analysis benefited from TMJ surgery.

Case Report

A twenty-eight year old white male presented to the Dental Facility at Abington Memorial Hospital with chief complaints of difficulty opening his mouth and chewing. In

addition, he had minimal dental work in the past due to limited mouth opening. He had been diagnosed with arthrogryposis multiplex congenita. There was significant physical involvement of the lower extremities as a result of his condition. His upper extremities appeared normal and well developed, and he was able to ambulate using bilateral crutches. The patient had dysarthric speech and compromised oral function, but his language function was intact. The patient was alert, coherent and cooperative. There was no complaint of specific pain. He had undergone numerous surgeries of the hip, knees and feet to correct and reduce disability over time. His dental history included orthodontic treatment and extraction of impacted bilateral maxillary first premolars, all four third molars, and a supernumerary tooth.

Radiographic findings from Magnetic Resonance Imaging showed menisci that were not well defined. The mandibular condyles were flattened indicating possible degenerative disease. Computed tomography showed an essentially normal appearance with no evidence of bone erosion. Electromyography of the left and right masseter and temporalis muscles was done. Both left and right masseters showed a severe muscle weakness pattern. The temporalis muscles showed moderate to severe muscle weakness patterns with the left side more affected than the right. The motor unit configuration was within normal limits for the size and morphology of the patient's muscles.

A surgical procedure was planned and eventually performed with the goal of increasing the range of motion of the mandible. The surgical treatment was accomplished under general anesthesia. There were no anesthetic complications. Bilateral intraoral temporomandibular joint coronoidectomies were performed followed by left and right menisectomies, lateral pterygoid

myotomies and capsular release via preauricular incisons.

Of significance during the examination procedure under open view, the condyle could not be moved to exit the fossa and was restricted entirely to rotational movement. This was even in the setting of the above mentioned surgical procedures. Initial maximum interincisal opening was measured at nine mm. upon presentation of the patient (Fig. 1.) Following bilateral coronoidectomy procedures, the maximum opening increased to fifteen mm. The final interincisal distance of twenty mm. (fig 2) was obtained after performing the TMJ arthrotomies.

Post-operatively, the patient was followed for eighteen weeks. Therapy consisted of a combination of home exercises and physical therapy sessions to maintain and increase mouth opening. The patient was instructed to use a Therabite® appliance, which enabled him to perform stretching exercises. At three weeks post-operatively, the patient was able to open sixteen mm. interincisally and could be pushed open to nineteen mm. With good compliance of exercise and physical therapy, his maximum range of motion increased to twenty mm. and he could be stretched to twenty-five mm. at ten weeks following surgery. At twelve weeks postoperatively, the patient was involved in a motor vehicle accident and consequently had discontinued physical therapy and home exercises. As a result there had been a significant decrease in mandibular opening range. At eighteen weeks post-operatively, the patient was able to open by himself to only eighteen mm. interincisally and was able to force open to twenty mm., which was still noted to be a significant improvement from pre-operative maximal opening. The patient, in addition, reported a marked improvement in speech and jaw function following this surgery and therapy. He continues to be followed in the out-patient clinic and has resumed use of

the Therabite® appliance.

Discussion

It has been suggested that the decision to utilize a surgical approach in treating patients with maxillofacial manifestations of Arthrogryposis should be approached with caution. Specifically, Epstein recommends that these patients only be considered for surgery if the cause of mandibular restriction is related to periarticular or intra-articular abnormalities.[1] Heffez states that surgery consisting of bilateral coronoidectomies would be beneficial only if the masseter and internal pterygoid muscles were not restricting the motion of the joint.[5] In this case, our patient's pre-operative condition was characterized by both rigid resistance to opening beyond nine millimeters interincisally and pronounced muscular weakness bilaterally. These conditions limited the patient's ability to masticate efficiently, and served as a hindrance to the administration of normal dental care. We felt that we could recommend surgery followed by an extended period of physical therapy, with the patient's understanding that the measurement of the success of this therapy would likely be subjective in it's significance to the patient. We were encouraged by the fact that despite his condition, this patient's upper limbs were functionally and anatomically normal, thus facilitating his ability to continue his own physical therapy at home postoperatively.

Surgical intra-operative findings were generally as anticipated; the mandibular opening increased modestly following the first surgical phase, which consisted of bilateral coronoidectomies. This finding is supported by Epstein, who demonstrated that

coronoidectomies alone did not significantly improve mandibular opening.[1] Both Heffez and Epstein suggest that alteration of the joint or structures surrounding the joint would be necessary in order to further improve opening.[1,5] Following coronoidectomies, bilateral modification to the TMJ's as described above yielded an additional six to seven millimeters of mandibular opening, providing a total interincisal distance of twenty millimeters in the immediate post-operative period. During the eighteen week period in which we have thus far followed this patient post-operatively, he has been able to maintain this distance, until recently, when he was forced to discontinue physical therapy and home excercises, resulting in an interincisal opening of eighteen millimeters. This decrease in opening following cessation of physical therapy is supported in the literature.[1,5,9] Notably, the patient and his father volunteered that post-operative subjective findings include both improved speech and efficiency of mastication. Additionally, more than doubling the patient's mandibular opening has certainly improved his ability to receive dental treatment of a more comprehensive nature.

Due to the complexity of AMC and its associated orofacial manifestations, we agree that careful screening prior to the decision to treat these cases surgically is definitely indicated. The necessitation of surgery should take into account clinical, radiographic, and EMG impressions, as well as the individual patient's age, quality of life, and expectations of surgical therapy.

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