Failure of Differentiation: Part II (Arthrogryposis, Camptodactyly, Clinodactyly, Madelung Deformity, Trigger Finger, and Trigger Thumb)

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• Arthrogryposis • Camptodactyly • Clinodactyly
• Madelung deformity • Trigger finger • Trigger thumb

ARTHROGRYPOSIS

The term "arthrogryposis" has traditionally been used to describe various conditions that present with congenital joint contractures.[1] Under this broad term, more than 150 separate disease entities that share elements of congenital contractures have been described.[2] The contractures are considered to be the end result of decreased intrauterine movement by the fetus after a period of normal development. This decreased movement may occur because of neuropathies, myopathies, abnormal connective tissue, or decreased intrauterine space.[2]

Classic arthrogryposis or arthrogryposis multiplex congenita describes a specific disease process. This term was first used by Stern[3] to describe three children in 1923. To add to the terminological confusion, Sheldon[4] gave a detailed clinical description of this same entity in 1932 and called it amyoplasia or amyoplasia congenita, emphasizing his belief that the disease was the result of aplasia or hypoplasia of certain muscle groups. This article focuses on the cause and upper extremity treatment considerations of this disorder, which is referred to as amyoplasia.

Distal arthrogryposis refers to a large subgroup of disorders in which the contractures primarily involve the hands and feet. Multiple classification schemes have been used to describe these conditions, but this group is extremely heterogenous and this term does not refer to a specific disease entity.[5–7]

Children who have amyoplasia present with a characteristic phenotype at birth. Most (84%) present with involvement of all four limbs, although

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11% present with only lower limb involvement and 5% with only upper limb involvement.[8] The limbs appear fusiform and cylindric with a pronounced decrease in muscle mass and a lack of flexion creases. There is decreased joint range of motion with a firm inelastic endpoint.[9] Sensation is normal and there are few associated visceral anomalies. The joint contractures are usually bilaterally symmetric. The shoulders are internally rotated and adducted, the elbows are extended, the wrists are flexed and ulnarly deviated, the thumb is adducted, and the fingers are partially flexed.[9] The hips are often flexed and abducted, and one or both hips are dislocated in one third of patients.[10] Knee flexion contractures are the most common presentation, but the child may present with knee extension contractures, and knee joint subluxation or dislocation may be present. Almost all children who have this condition have rigid clubfeet, although congenital vertical talus or another foot deformity may be present. Congenital scoliosis is unusual and should trigger the work-up of other neuromuscular disorders. Neuromuscular scoliosis may develop in 30% of patients, however.[10] Some 90% of patients who have all four limbs involved have a characteristic frontal midline capillary hemangioma.[11]

Amyoplasia is sporadic, with an incidence of 1 of every 10,000 live births.[12] Fifteen identical twin cohorts have been reported in the literature as being discordant for the disease.[13] This finding strongly suggests that genetics do not play a major role in the disease, and that some factors in twins may contribute to the condition.[13] Several authors have primarily attributed amyoplasia to be the end result of damage to the anterior horn cells of the spinal cord.[14,15] Others believe that a primary myogenic form may exist.[12] Multiple studies have not found a clear environmental cause.[16]

Several authors have commented that children who have amyoplasia are often of above-average intelligence and have the potential to become independent adults despite significant physical limitations.[9,17] Bevan and colleagues[18] suggest that to help a child who has amyoplasia reach this goal, treatment should be focused on communication, activities of daily living, mobilization, and ambulation, in decreasing order of importance. With this in mind, treatment of the upper extremity must focus on preserving or increasing motion and allowing the upper extremities to be positioned at the tabletop level for self-care and access to computers.[17] Because children who have amyoplasia are extremely limited with regard to upper extremity strength, they depend on bimanual patterns and this must be preserved.[17] Van Heest and colleagues[19] reported one patient who had a loss in function after the development of an elbow flexion contracture. This condition, combined with the patient's pre-existing contralateral elbow extension contracture, led to a decline in the ability to perform activities of daily living. Occupational therapy is often helpful in assisting with learning adaptive skills and providing assistive equipment.

Most frequently, the child who has upper extremity involvement benefits from treatment of elbow and wrist contractures. The typical arthrogrypotic deformity of the elbows is full extension with some triceps function.[17] The lack of passive elbow flexion is particularly disabling because it does not allow hand-to-mouth function. Children who have amyoplasia who have passive elbow flexion but lack active flexion are often able to use strategies, such as trunk swaying or table top propping, to flex the elbow for self-feeding.[20] Tendon transfers to restore active flexion of the elbow are difficult given the relative lack of donor musculature and carry a significant risk for creating an elbow flexion contracture. They seem to provide little additional functional benefit for most children who have amyoplasia.[20]

Initial treatment of the upper extremities in the infant who has amyoplasia begins with gentle manipulation and splinting. This treatment may be particularly helpful with regard to the wrist flexion and elbow extension contractures. Therapy may also assist in maintaining muscle mass and strength in the child who has amyoplasia.[21] Smith and Drennan[22] evaluated 17 infants who had wrist flexion contractures and found that patients who had distal arthrogryposis were more responsive to early casting, serial casting, and orthotics than patients who had amyoplasia.

Timing of upper extremity surgery is controversial. Mennen advocated an early one-stage corrective procedure between 3 and 6 months of age.[23] He recommended proximal row carpectomy to address the wrist flexion contracture and the triceps-to-radius transfer to achieve active elbow flexion. In his report on 47 limbs, he found that patients who were operated on early gained more active motion and had improved carpal bone remodeling. He also believed that the repositioning the wrist allowed for improved finger function and noted that creases developed over the finger joints after wrist surgery.

Because amyoplasia frequently involves all four extremities, however, some advocate waiting until the child begins to ambulate before beginning upper extremity surgery. This recommendation may result in a significant time delay, given that clubfeet, knee contractures, and hip dislocations often need to be addressed. In the nonambulatory