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Classic Arthrogyrposis (Sporadic Amyoplasia)

Introduction

Arthrogryposis is a pediatric syndrome associated with over 400 specific neuromuscular or genetic diagnoses.[1] Children with arthrogryposis are born with multiple non-progressive joint contractures.[1,2] Arthrogryposis was historically considered a medical diagnosis but due to recent advances in genetics and pathophysiology, the presence of multiple contractures at birth is now considered a clinical sign of an associated diagnosis.[1] The clinical finding of arthrogryposis necessitates an extensive medical differential diagnosis to provide families with appropriate genetic counseling and determine appropriate management. Hall proposed three sub-categories of arthrogryposis employed to aid with differential diagnosis. [1,3] The sub-categories are limbs only, limbs plus other systems, neuromuscular involvement with central neurologic or intellectual dysfunction.[1] The classic form of arthrogryposis is sporadic amyoplasia, or "lack of muscle development".[4,5] Thirty percent of children with arthrogryposis are eventually diagnosed with sporadic amyoplasia.[4] Overall incidence of sporadic amyoplasia is 1 out of 10,000 live births.[2] Amyoplasia can be further broken down into one of five sub-types: four-limb symmetric, three-limb, upper limbs only, lower limbs only or severe involvement.[5] The classic arthrogryposis diagnosis is sporadic amyoplasia and involves contractures of all four limbs.[4,5] The purpose of this paper is to describe the clinical manifestation, etiology, pathophysiology, and physical therapy management of children with classic arthrogryposis or amyoplasia within the context of the ICF.

Clinical Presentation:

Figure 1 depicts the typical presentation of an infant with sporadic amyoplasia affecting all four limbs.[6] Upper extremities contractures involve shoulder internal rotation, elbow extension, forearm pronation, wrist flexion and camptodactyly.[5] Flexion contractures predominate in the hips and knees with hip abduction, hip external rotation and equinovarus of the feet. Lower extremity contractures vary and may present instead with knee extension contractures or dislocated hips.[5] All four limbs are short in stature with decreased muscle mass, dimpling of involved joints and decreased flexion creases.[5] A more severe and less common form of amyoplasia also involves all of the extremities but in contrast presents with elbow flexion, possible scoliosis, hip flexion, knee flexion, and more pronounced equinovarus.[5] Children with amyoplasia also have an increased incidence of gastroschis and/or bowel atresia.[5] Although not specifically studied, intelligence is normal and newborns are alert.[5] Range of motion is most limited at birth and typically improves with appropriate treatment. The degree of improvement is dependent upon initial severity and proper medical management including surgery and therapy.[2,5] However, long term function is more related to social and environmental factors such as family support, patient personality, education and strategies encouraging early independence.[2]

Etiology and Pathogenesis:

The main causative factor in the development of amyoplasia and all forms of arthrogryposis is fetal akinesia or decreased movement in utero. The etiology and pathophysiology responsible for reduced movement is heterogeneous and lacks a definitive pre-natal predicator.[1] More severe contractures at birth are related to an earlier onset of decreased fetal movement.[5] Table 1 outlines the implicated intrinsic, extrinsic and environmental factors associated with the development of fetal akinesia resulting in amyoplasia. More recent studies propose that fetal akinesia is caused by decreased fetal blood flow occurring between the seventh to twelfth weeks of gestation.[5] Hypotension may damage the development and maturation of anterior horn cells resulting in disruption of muscle development and function thereby reducing fetal movements promoting and disuse atrophy of involved muscles supplied by affected motor units. Hall hypothesizes,

"Amyoplasia is not a primary malformation, but rather a fetal process involving interruption in normal development with an overlay of a whole series of vascular compromises or tissue disruptions which then leads to subsequent loss of normal tissue and transformation of other tissues back to more primitive or pluripotential state. Then fetal akinesia develops with all its secondary effects."[5]

Extremity muscle mass is reduced in children with amyoplasia and biopsies show increased fatty-fibrinous deposits interspersed with normal muscle cells within the same muscle. Changes within the muscle reduce movement capacity and also leads to disuse atrophy further reducing the movement and strength. Although joints initially develop normally lack of fetal movement results in collagenesis around the joints further restricting motion. Lack of movement creates a cascade of events affecting normal development.

Environmental	Intrinsic	Extrinsic
Maternal Chronic Illness (Diabetes, Multiple Sclerosis, dystrophies, myasthenia gravis)	Myopathy	Uterine Septum
Maternal Infection with hyperthermia	Damage to peripheral and or central nervous system	Trauma
Reduced Blood Supply	Neurogenic Damage	Multiple Pregnancy (twins, triplets etc.)
First Trimester Pregnancy Complications	Connective tissue disorder	
	Genetic Mutation	

Table 1. *Potential Causes Implicated in Amyoplasia*[1,5]

Physical Therapy Management

The primary body structure/function impairment for children with sporadic arthrogryposis is decreased joint range of motion and muscle strength. [7] (See table 2) Physical therapy treatments focus on improving joint range of motion, extremity strength, core strength, and postural control utilizing age appropriate developmental and task specific activities.[7] Appropriate adaptive equipment and bracing

recommendations are also necessary to maximize the child's functional activities and participation in age appropriate and meaningful activities.[7] Families are initially educated on proper stretching, positioning and splint use of the both the upper and lower extremities.[8,9] Involved joints typically demonstrate a firm and end-feel to restricted motions.[2] In the upper extremity elbow flexion, shoulder external rotation, shoulder elevation, wrist extension, forearm supination and finger extension is limited. The lower extremities exhibit reduced hip adduction, internal rotation, knee extension, ankle dorsiflexion and eversion. Parents are educated to perform stretching 3-4 times a day holding 20-30 seconds.[7,10] Bath time and diaper changes are excellent reminders for PROM. Palmer et al. found that passive range of motion was effective to improve range of motion in affected joints and advocates frequent therapy sessions to promote parental independence with stretching.[10] Once a desired range of motion is achieved splints are applied to maintain the new range. The foot and ankle complex is the most frequently involved joint in all types of arthrogryposis. [7] Contractures vary in severity from club feet to mild equinovarus. The ROM goal is to reach at least 0 degrees of dorsiflexion with neutral calcaneal position to promote development of standing and ambulation. Range of motion is achieved through stretching, serial casts (Ponsetti Method), ankle foot orthoses and/or surgery. [3,7,10,11] The Ponsetti method has been found to be effective treatment for club feet especially when initiated within the first month of life for children with arthrogryposis.[11] Although splinting and/or serial casting are necessary to improve ROM Kroksmark et al. warn that excessive use of immobilization may decrease opportunities for strengthening and active range of motion.[12] Kroksmark et al. found that a reduction in knee flexion contractures and increased muscle strength was more associated with ambulation than range of motion.[12] One avenue may be to use long static stretching at night and standers during the day and encourage active movements during facilitated task specific play and daily routines. For example, a child with knee flexion contractures is fit for a snug seat stander that allows for a comfortable stretch into knee extension while child plays with toys that encourage elbow flexion and reaching.[13,14] Standing is encouraged for at least one hour a day to improve lower extremity range of motion.[13] At night serial casts or splinting for knee extension are applied for a long static stretch.

Most children with amyoplasia progress to ambulation by the age of 6 with or without orthotics and assistive devices.[4] Ambulation by the age of 24 months is related to progression to community ambulation.[12] Rolling walkers or gait trainers along with bracing, such as KAFOs or AFOs, may be necessary to achieve independent ambulation. The physical therapist must assess lower extremity and upper extremity strength and determine which device is most appropriate. Gait trainers are most appropriate for children with limited ability to support his/her weight with upper extremities and significant lower extremity weakness especially of hip and knee extensors.[7] For children with reduced grasp or upper extremity strength, forearm supports or custom molded upper adaptations can be added to rolling walkers.[7] KAFOs may be necessary for children with weak quadriceps and gluteals that are not responding to strengthening. Locked KAFOs increase stability for ambulation but are cumbersome, reduce speed increase energy expenditure of gait, and are associated with household ambulation. The least amount of lower extremity bracing is desired for progression to community ambulation and to minimize the energy expenditure of gait.[15] Strength of hip and knee extension is related to functional ambulation for children with amyoplasia.[12] Sit to stand activities and games are incorporated into therapy, play and child's daily for strengthening. Families are also encouraged to place their child in

supported or unsupported standing with and without assistive devices for task specific training during typical daily routines such as dressing, brushing their teeth or reaching.

Sporadic Amyoplasia (Classic Arthrogryposis)

Body Structure/ Function	Activity Limitations	Participation Restrictions
•Limited Active and/or Passive Joint Range of motion •Reduced Strength of involved muscles •Joint surfaces are "squared", lacking normal rounded shape •Decreased Deep Tendon Reflexes •Decreased postural control in sitting and standing •Hip Dislocation (30%) •Scoliosis (30%) •Gastrointestinal Dysfunction •Normal Sensation	•Gross and Fine Motor Delays •Feeding Difficulties •Difficulties with reach, grasp and manipulation with hands •Reduced ambulation due to increased energy expenditure •Dependent transfers •Difficulties with ADLs ex: dressing, eating, bathing, toileting, grooming	•Reduced Play with peers •Reduced Recreation Involvement •Unable to keep up with peers in school •Unable to participate in community events or activities (park, shopping mall, kids museums, bike riding)

Personal/Environmental Factors

- Third Party Payer may restrict purchase of adpated equipment
- Time demands on family
- Home may not be accessible
- Education of parents
- Financial/emotional stress on families
- Access to specialized clinics or health care

Table 2: *ICF Considerations for Children with Sporadic Amyoplasia*[2-4,7,8]

Community Resources and Education for Parents of Children with Sporadic Amyoplasia

As stated earlier, functional outcomes for children with amyoplasia are related to personal and environmental factors impacting independent behaviors more than range of motion alone. Therefore, one of the most important roles of the physical therapist is to educate families on how to support and encourage independent function, participation and physical activity for their children by recommending necessary task and environment modifications. For example, if a child cannot walk long distances a

power or manual wheelchair may be recommended to encourage independent mobility in the community instead of a pediatric stroller. On-line support groups, national associations and Facebook pages are an excellent way to meet other families and share ideas and encouragement.[3,16,17] National conferences, seminars and mini-meet ups are offered by the Arthrogryposis Multiplex Congentia Support Inc. (AMC).[16] Families may gain additional Insight and wider perspective through discussions with families who have already lived through a similar situation. Therapists can provide information on adapted equipment, toys, developmentally appropriate activities to encourage gross motor development, and community recreational activities. Many communities offer adapted sports, art classes, adapted bicycles, and swim lessons. For example, Greenville Recreation and Parks offer adapted baseball, football and swim programs.[18,19] A local chapter of National AMBUCS provides free adapted bicycles for children with disabilities.[20] Adapted bicycles are individually modifiable for optimal use of upper extremity only, lower extremity only or a combination of a UE and LE drive.[21] Lastly, aquatic programs are also an effective, fun and motivating exercise and recreation and social outlet for children with disabilities similar to amyoplasia.[22-24]