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RESEARCH REVIEW

Rehabilitation across the lifespan for individuals with arthrogryposis

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Abstract

Arthrogryposis multiplex congenita (AMC) can be a perplexing diagnosis that consists of limited range of motion (ROM) and decreased muscle strength in multiple joints. The person with AMC often possesses a certain tenacity and "spunk" that assists them with adjusting and adapting to the realities of daily life. The rehabilitation process assists the individual with AMC in achieving and maintaining the maximal active and passive range of motion and strength in order to participate in activities of daily living (ADL) throughout the developmental stages. The result of this life-long process is greatly impacted by collaboration among the multidisciplinary teams. Ultimately, rehabilitation should focus on three levels of treatment: (a) body structure, (b) activity, and (c) participation. This article describes rehabilitation across the lifespan—focusing on the therapeutic needs in the infant, toddler, school age and teenage/adult years—while also highlighting opportunities for improvement.

KEYWORDS

activities of daily living, arthrogryposis multiplex congenita, orthotics, range of motion, rehabilitation

1 | INTRODUCTION

Rehabilitation often refers to seeking or regaining function affected by a medical condition or an injury. For the person with a congenital condition such as arthrogryposis multiplex congenita (AMC), rehabilitation does not "regain function" but seeks to gain function throughout the developmental process. Intervention maximizes the abilities a child may have to provide for the best outcomes throughout the lifespan. In general, AMC can be a perplexing diagnosis of limited range of motion (ROM) and decreased muscle strength in multiple joints (Kowalczyk & Felus, 2016). The individual with AMC, however, often possesses a certain tenacity and "spunk" that assists them with adjusting to these limitations. The rehabilitation process assists with achieving and maintaining the maximal ROM and strength in order to participate in activities of daily living (ADL). Rehabilitation should focus on three levels of treatment: (a) body structure—the anatomical parts of the body, (b) activity—the execution of a task or action by an individual, and (c) participation—the involvement in a life situation (World Health Organization, 2001). For the purpose of this article, the body structure component describes the anatomical components such as ROM and orthotics; the activity and participation domains grouped together describe the functional activities typically addressed in each of the stages—infant, toddler, school age, and teenage/adult years. Opportunities for growth for Health Care Professionals (HCP) are identified corporately.

To effectively impact the quality of life of a child with AMC, rehabilitation should be early, comprehensive, and multidisciplinary (Binkiewicz-Glinska et al., 2013). Multidisciplinary team members include the core family and multiple HCP depending on the severity of AMC (Figure 1). The child and caregivers benefit from family-focused care, repeated education, and reinforcement at each consecutive treatment stage.

2 | INFANT STAGE (0–18 MONTHS)

The infant stage begins at birth with the presentation of contractures and depending on the type of AMC multiple comorbidities such as hernias, micrognathia, severe scoliosis, and microcephaly (Hall, 2014). Medical stability is the priority; however, therapeutic approaches should begin as soon as possible. As shown in Table 1, early therapeutic intervention focuses on improving passive and active ROM in all affected joints, facilitating active motion demonstrated by antigravity strength, and correction of fixed deformities that affect daily living skills (Kowalczyk & Felus, 2016). The infant with AMC may receive therapy in various environments such as the neonatal intensive care unit (NICU), daycare, clinic, and home. Depending on the setting and the learning styles of the caregivers, health care professionals (HCP) must embrace different education methods, with the potential use of audio-visual resources and social media to provide an alternate avenue for knowledge propagation.

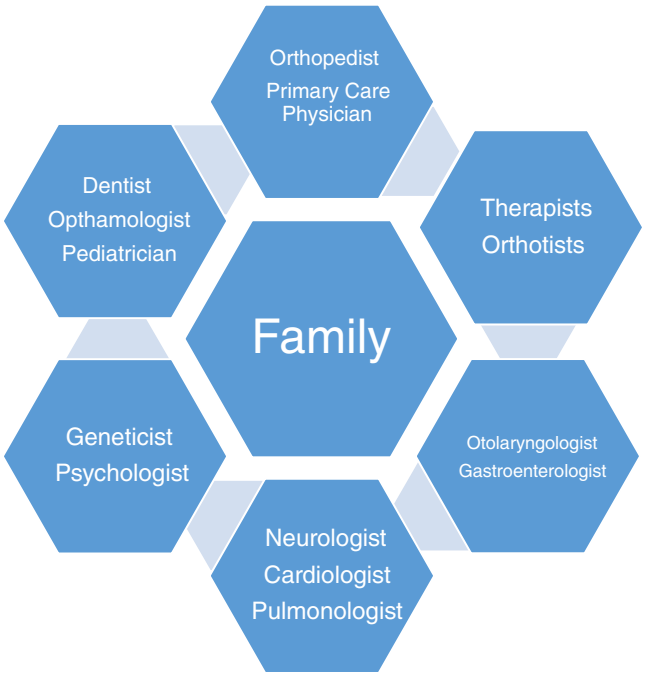

FIGURE 1 Multidisciplinary team members. Figure depicting potential team members for AMC patients. AMC, arthrogryposis multiplex congenita

2.1 | Body structure

Connective tissues are most compliant shortly after birth and early manipulation may provide an opportunity for a somewhat "typical" joint to form (van Bosse et al., 2017). Because the anatomical alignment is often a challenge to discern, gentle passive ROM is performed with mindfulness of optimal alignment. Gentle handling can be completed with the head and neck if limited cervical range prevents midline head alignment (Kaplan, Coulter, & Fetters, 2013). Due to the cephalocaudal motor development of an infant, a lack of neutral head positioning impacts vision, feeding, and body movement (Alexander, Boehme, & Cupps, 1993). Management strategies promoting an increased range of motion and joint stability for the upper and lower extremities are determined after careful analysis of the limiting factors in all joints. Custom molded low-temperature orthotics, casting, and taping provide a constant or dynamic stretch to the muscle and joints. Clubfeet, described as more severe and rigid than idiopathic clubfeet, are the most frequent foot deformities in infants with AMC (Carlson, Speck, Vicari, & Wenger, 1985). Conservative management of clubfeet must be initiated as early as possible in preparation for proper positioning on wheelchair footrests, standing, and/or gait. Orthotics used after clubfoot correction may vary from those of the traditional Ponseti method. The most common

TABLE 1 Rehabilitation general Review[a]

	Infant	Toddler	School age	Teenage/adult
Therapy	Multiple environments High intensity	Multiple environments High intensity	School environment Episodic care	Minimal formal services
Body function	A/PROM[b] Activation of movements Correction of deformities Orthotics/taping	A/PROM Strengthening Postoperative protocols Orthotics	A/PROM Strengthening Postoperative protocols Orthotics	A/PROM and strengthening through "life" Healthy lifestyle-exercise, cardio vascular, minimizing weight gain
Activity/ participation	Floor mobility-transitional movements; rolling, sitting Fine motor-exploratory play reaching, grasping, holding	Mobility-transitional movements; standing/gait Fine motor-explorative play ADL-feeding, emerging community participant	Mobility-school accessibility Fine motor- Assistive technology ADL-autonomy from home caregiver, increased community participation	Mobility-full accessibility within environment/community Fine motor-job related IADL[c] skills

Abbreviation: AMC, arthrogryposis multiplex congenita.
[a]Summary table of therapy settings, body functions, and levels of activity/community participation across the lifespan of AMC patients.
[b]Active/passive range of motion.
[c]Instrumental activities of daily living.

orthoses, fabricated from high temperature, lightweight thermoplastic material, used during the first year of life for the lower extremities are ankle-foot orthosis (AFO), knee orthosis (KO), or knee-ankle-foot orthosis (KAFO). Knee alignment in an infant with AMC varies greatly in the sagittal, frontal, and transverse planes. Due to this, the orthoses may be configured without knee or ankle joints (Palmer, MacEwen, Bowen, & Mathews, 1985). In regards to the upper extremity, orthoses fabricated to address the varying joint contractures of the elbow, wrist and fingers require frequent monitoring to advance the gained ROM. Surgical interventions at any joint level are considered from initial infancy throughout the ages and stages, followed by variable therapeutic protocols necessary to promote joint alignment and functional performance. The multidisciplinary team is essential as surgeons, therapists, and orthotists work together to combat recurrence, minimize surgical interventions, and maximize the patient's outcome.

As ROM increases, an infant may also gain selective muscle strength. Kroksmark, Kimber, Jerre, Beckung, and Tulinius (2006) reported a correlation between muscle strength and motor function. They recommended developing muscle strength by early stimulation of active movements and minimizing immobilization. Therapists promote ROM and muscle activation through "typical" developmental postures. For example, in prone the infant gains antigravity strength for head control, while simultaneously promoting increased elbow flexion for hand-to-mouth activities.

2.2 | Activity/participation

Mobility enables a child to explore and interact with their environment, which is crucial for cognitive development (Alexander et al., 1993). The therapist's role is to guide the acquisition of these skills while promoting "typical" developmental postures. It is more important that a child *does* explore their environment rather than focusing on *how* they explore their environment. Infants need to be successful at mobility so they will continue to move. A systematic review of typically developing children ages birth to 5-years-old reported those with increased higher duration and/or frequency of physical activity had statistically significant ($p < .05$) beneficial effects on at least one cognitive domain (Carson et al., 2016).

For many children with AMC, rolling is the initial method for mobility. Some infants develop a side-lying scoot using excessive trunk flexion and extension to move across a room. Infants with AMC seldom develop a traditional quadruped or army crawl. Sitting is a critical developmental skill as it influences upper extremity function, allows upright functional self-care skills, and allows the child to refine cognitive, perceptual, and social skills (Banas & Gorgon, 2014). For children with AMC, the actual hip and knee contractures may preclude typical positioning and require adapted seating systems. In the sitting position, the infant has the opportunity to increase upper extremity function/awareness. Upright positioning can facilitate arms coming to the midline, while the use of a tray provides propping opportunities for passive elbow flexion to aid the developmental skills of exploring hand-to-mouth or manipulative play. Visual skills and early social skills including interaction with others occurs when sitting upright. Explorative play includes the development of early self-feeding skills, such as the attempt for hands to bring a small piece of food from tray to mouth. Facilitation of transitional skills in and out of a seated position are imperative. In AMC, upper extremity contractures and weakness may interfere with this transitional skill of assuming a sitting position from the floor. The infant often adapts and utilizes the core muscles and head control to attain upright sitting. Upright floor mobility often occurs once in the sitting position with the bottom scoot technique, utilizing body momentum to accomplish access to the environment. For those with wrist flexion contractures, weight bearing through the dorsum of the wrist assists with scooting.

Development of upper extremity skills progresses along with gross motor skills. Caregivers and therapists need to discover developmentally appropriate toys an infant can reach for, grasp, and manipulate. Techniques to stimulate wrist and finger movement can include ROM, taping, splinting, or sensory stimulation. Affording opportunities for hand-to-mouth through passive or active movements provides oral motor stimulation, can be a method for self-organization or calming, and is a precursor for self-feeding.

Following typical developmental motor acquisition patterns occurring at the end of the first year, therapists may introduce preliminary standing. In a systematic review of children with motor impairments, Paleg, Smith, and Glickman (2013) recommended the use of standing devices to positively affect bone mineral density and to improve lower extremity range of motion. Options for supported standing include standing frames and/or orthotics. If orthotic bracing is required, functional antigravity strength will be assessed to determine the optimal design of the orthotic. The older infant will use strong core muscles, learning to find their center of gravity to balance over the base of support. With the help of custom molded orthotics, the older infant will have the foundation to succeed with balanced standing and future walking (van Bosse et al., 2017).

3 | TODDLER STAGE (18 MONTHS–4 YEARS OF AGE)

The toddler with AMC often spends time learning and exploring to gain skills that will translate to greater independence. Limited mobility and difficulty accessing objects in their environment create challenges in exploration. Therapeutic settings at this age may vary from home, daycare, and/or therapy gyms and settings. In order to qualify for intervention services, standardized evaluation tools may be used to compare the child with AMC with a typically developing child. Play and exploration in the toddler years builds strength and confidence in ADLs needed for the future. Increasing experiences for mobility, balance, and fine motor activities enhances opportunities for independence (Table 1).

3.1 | Body structure

The peak growth periods associated with the toddler age increases limb length, which both enables and challenges function. Overall, the need to work on ROM and strengthening exercises remains constant.

Surgical intervention occurring in the toddler stage addresses limits in body structure; protocol exercises are imperative to maximize outcomes and create optimal functional alignment (Abdel-Ghani, Mahmoud, Shaheen, & Abdel-Wahed, 2017; Lake & Oishi, 2015; Van Heest, James, Lewica, & Anderson, 2008). Orthotics introduced in the infant stage continue to support, facilitate, and provide stabilization for passive and active ROM. Some children with AMC do not have sufficient strength to lift their arms against gravity. These children may discover, on their own, alternative leverage techniques that do not require assistive devices. They may find the use of overhead slings or upper extremity exoskeletons (Figure 2) helpful in gaining antigravity use of the upper body and subsequently promoting fine motor skills (Babik et al., 2016).

3.2 | Activity/participation

Initial mobility developed in the infant stage continues into the toddler stage. The ability to move independently to access objects facilitates independence and problem solving. As mentioned earlier, the key is not to focus on how the child gets from point A to point B, but rather celebrate the ability. The wiggle, scoot, and roll at this stage forms the foundational movement skills needed for future activities of daily living.

During the toddler stage, floor mobility may evolve into the ability to stand and walk. Transitional movement skills may progress to learning how to assume standing up from a chair or even from the floor. Assistive devices, such as a stander or gait trainer, continue to be helpful in gaining upright skills as well as helping with the management of lower extremity joint contractures (Gmelig Meyling, Ketelaar, Kuijper, Voorman, & Buizer, 2018; Sival, Prechtl, Sonder, & Touwen, 1993). While most children with AMC will have a level of standing and stepping ability by the end of preschool, not all will walk in adulthood (Donohoe, Pruszcynski, Rogers, & Bowen, 2019). The use of gait training devices can prompt stepping skills with and without orthotics, which leads to improved walking skills, increased exploration, and exercise. Participation in the toddler stage includes peer and/or family interactions occurring in multiple environments such as preschool, community libraries, and outdoor playgrounds. Hands-free gait trainers do not necessarily allow better long-term ambulation but do permit better exploration with peers (McKeever, Rossen, Scott, Robinson-Vincent, & Wright, 2013). As children develop adequate balance to stand and step, a walker may still be necessary to prevent falling. Posterior walkers allow for greater hip and trunk extension and improve energy conservation when compared to anterior walkers (Park, Park, & Kim, 2001); however, a therapist should always individualize treatment to the specific child. Along with walking strategies, toddlers must learn the sensation of falling and the strategies to recover balance in order to become independent ambulators. Additional challenges occur with environmental changes such as ambulating on uneven or outdoor terrain including grass, gravel, or mulch and elevation changes such as stairs or curbs.

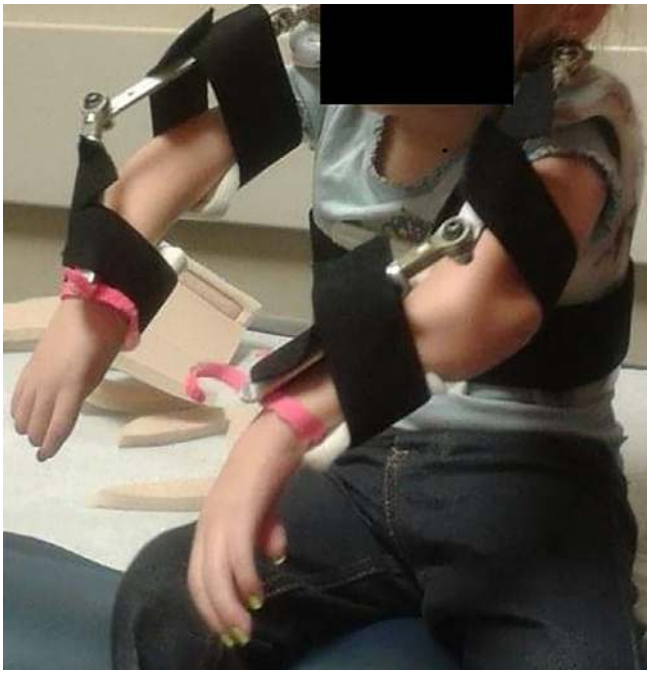

FIGURE 2 Exoskeleton example. Photograph illustrating upper extremity exoskeleton

Many toddlers with AMC continue to wear orthotics. A hip knee ankle foot orthotic (HKAFO) with a mechanical hip joint may be used to stabilize the hips during walking. Typically, KAFOs with an open knee joint are used when coronal and transverse plane knee stability is required (Bartonek, 2015; Eriksson, Gutierrez-Farewik, Brostrom, & Bartonek, 2010). For children with knee extensor weakness, the KAFO knee joint can be locked for stability in the sagittal plane (Donohoe, 2006; Eriksson et al., 2010; Graubert, Chaplin, & Jaffe, 1998; Staheli, Hall, Jaffe, & Paholke, 1998).

As balance improves, reduction of support occurs with less stabilization of the hips and knees. Solid AFOs are commonly used to stabilize the foot and ankle to provide a stable platform for standing and walking and help prevent clubfoot recurrence (Bartonek, 2015; Donohoe, 2006; Eriksson et al., 2010; Graubert et al., 1998; Staheli et al., 1998). The use of hinged (articulated) AFOs with limited range of motion have their role as they provide increased mobility for functional tasks (Bartonek, 2015; Eriksson et al., 2010). For those with plantarflexion weakness, a carbon fiber spring ankle joint can improve kinematic and kinetic gait parameters, stride length, and walking speed (Bartonek, Eriksson, & Gutierrez-Farewik, 2007).

Explorative play with toys that require manipulation is integral for fine motor development. Although electronic devices that require a tap or a swipe allow easy access to play, they do not allow for the practice of fine motor skills needed for feeding, dressing, bathing, and handwriting. It is important for the child with AMC to have the opportunity to pinch, pull, hold, and rotate. Adaptations and modifications may be provided to enhance this participation. The strategies needed for this play will translate to strategies required for ADLs in the future.

Self-feeding skills develop during this age. Anti-gravity strength, alignment of the upper extremity, and willingness to use adaptive equipment can affect this skill. Children with AMC who cannot bring

their hand-to-mouth often cannot self-feed like a typical toddler. Use of tools with adapted handles may support independence. Younger children may eat straight off the plate when tools are too difficult. Although helpful for independence, this practice becomes socially unacceptable at older ages; therefore, therapy should understand the child's satisfaction with independence while still introducing methods for typical feeding.

4 | SCHOOL AGE STAGE (5–12 YEARS OF AGE)

During the school age years, the child develops increased autonomy as they participate in school environments and develop outside friendships. Children assume greater responsibility in their own care and participation in therapy. Staheli et al. (1998) reported that while the self-care domain is the priority, facilitating school-oriented tasks like writing, drawing, and painting are also necessary. Therapy focuses on the domains of self-care and educational participation with adaptations, modifications, and alternative approaches used to achieve maximal ADL independence (Table 1).

Therapy tends to be episodic during this stage. Families present to therapy with specific goals. The HCPs evaluate the child using standardized assessments and observations of functional capacity and assess ROM and strength to determine sufficiency for performing tasks. Together, the families and the HCP formulate a plan to address ROM, strength, and endurance to accomplish activities for greater participation in all areas of life.

4.1 | Body structure

Although the primary emphasis at this age shifts from daily ROM and stretching exercises to ADL independence, ROM and stretching remain important (Palmer et al., 1985; Vermaak, 2012). Due to the risk of joint stiffness and contracture recurrence, ROM and stretching is a lifelong responsibility for the patient with AMC. Nighttime splints may be necessary to maintain ROM.

A school-aged child has the ability to provide input and opinion to a plan of care. An older child's increased awareness and compliance with instructions and precautions makes the timing of various surgical interventions more appropriate. The multidisciplinary team may consider muscle transfers or rotational osteotomies to improve joint alignment and increase active functional motion. Postoperative therapy focusses on specific protocols for a given surgery to facilitate functional improvement. Due to the frequent use of compensatory muscle strategies, continued therapeutic muscle strengthening is beneficial for children with AMC.

4.2 | Activity/participation

As the child begins school and spends time away from home and family, transfer skills, and walking become highly important (Dillon, Bjornson, Jaffe, Hall, & Song, 2009; Sells, Jaffe, & Hall, 1996). Therapeutic intervention for the walking child focuses on speed, terrain, endurance, and prevention of falling. Walking patterns for ambulatory individuals with AMC are established in terms of step length, truncal movement, and hip strength (Bohm, Dussa, Multerer, & Doderlein, 2013; Stief, Bohm, Ebert, Doderlein, & Meurer, 2014). Therapy for the non-ambulatory child focuses on the appropriate manual or power wheelchair and the ability to maneuver various distances and terrains. Environmental adaptations facilitate transfer and mobility skills within the school environment.

Indications for specific types of braces are identical to those of the toddler stage. Safety is an important concern, especially in a school or community setting with multiple peers and an increased risk of falls. Assistive devices such as canes or walkers may be necessary to increase stability and safety. For children with limited walking ability, wheelchairs are an alternative to facilitate transportation. Wheelchairs allow for faster access to the environment, cause less fatigue than ambulation, and decrease the risk of falls thus leading to increased school participation and social interaction with peers.

During the schoolday, primary caregivers are no longer present to assist with daily routines such as eating with peers in the cafeteria and toileting during classroom breaks. Increased autonomy in all aspects of self-care is imperative. Although the sample size was small, Joubert and Franzsen (2015) reported that children with AMC do not achieve independence in their self-care skills at the same rate as the normative population. A therapist must understand the limitations of strength and ROM and provide adapted equipment, teach modified positions, and/or advocate assertiveness on behalf of the child in order to obtain maximal independence.

Children with AMC need to be a part of their classroom. Handwriting skills and computer skills are necessary for participation in group centers and class projects. As with all school age children, involvement in play and leisure activities with peers has increasing importance. Exploration of both educational and vocational avenues in areas such as science, art, and music are more prominent (Staheli et al., 1998). Therapeutic intervention should facilitate maximum independence and participation through alternative approaches, environmental adaptations, equipment, and/or assistive devices (Joubert & Franzsen, 2015).

5 | TEENAGE/ADULT YEARS (13 YEARS AND UP)

Teens and adults are busy “doing life.” Basic ADL and mobility skills are functional within their family routine. As early teens transition to adulthood the focus shift toward increased independence, education, and employment with less emphasis on physicality. Access to rehabilitation services declines after 18 years of age, with limited resources available to the adult AMC population (Table 1).

5.1 | Body structure

Although little time and energy is left to continue the recommended therapy regimens, ongoing exercise as part of a healthy lifestyle will help maintain ROM and mobility skills that were a critical component

of therapy during childhood. Muscle stretching and strengthening occurs during the tasks of daily living. The use of orthoses for walking varies from person to person depending on needs and preferences (Eriksson, Jylli, Villard, Kroksmark, & Bartonek, 2018).

Previous research by Nouraei, Sawatzky, MacGillivray, and Hall (2017) showed that adults with AMC are much less physically active compared to the able-bodied population based on metabolic equivalent units (13 vs. 22 metabolic equivalent units). This study did not report a greater prevalence of obesity in adults with AMC compared to the general population. Nevertheless, regular exercise helps maintain a healthy weight, which plays a role in minimizing joint deterioration associated with age. Maintaining a healthy weight allows less energy to be put into ADLs. This allows more vitality for participation in social activities, employment, and family life, which is an important consideration as almost half of adults with AMC live with a spouse or partner (Nouraei et al., 2017). Similar to the general population, adults with AMC need regular follow up with a general practitioner to monitor overall health and screen for medical issues such as heart disease and diabetes (Centers for Disease Control, 2018).

5.2 | Activity/participation

Rehabilitation in the broader sense is still important. Activities required for school or work include environmental participation such as transferring to and from public transportation, or for living alone such as doing laundry and cooking. To improve a person's capability to participate in more activities, a continual focus on a healthy lifestyle including good nutrition and exercise is essential.

Keeping the body fit with cardiovascular and strength training furthers one's ability to carry out ADLs such as wheelchair transfers, cooking, dressing, family life, and work life. It is now understood that one third of people with AMC have a type called Amyoplasia (Hall, 2014). This is a condition whereby the anterior horn cells in the spinal cord are affected at about 8 to 10 weeks in-utero, resulting in muscle weakness and contractures that are so prevalent in these newborns (Hall, 2014). Thus, some similarities exist between AMC and patients with spinal cord injury or disease (SCI/SCD, respectively). Because there are so few guidelines for exercise for people with disabilities in general, we need to look to other population groups to find potentially suitable guidelines. For rehabilitation purposes, it is reasonable to extrapolate guidelines formed for the SCI/SCD populations to Amyoplasia and potentially other AMC patients. The latest physical fitness guidelines from Martin Ginis et al. (2018) suggest moderate or vigorous strength training twice per week and 20 minutes of aerobic exercise twice per week. If walking or cycling is difficult due to joint limitations, water-based exercise like swimming or water aerobics is a good substitute. However, one must be mindful that changing into and out of swim attire and showering in public facilities may present a challenge to many with AMC. Rowing is another alternative. Steen, Christensen, and Samargian (2017) indicated that 64% of their 22 adults with Amyoplasia (20 to 91 years of age) were actively engaged in a variety of exercise activities including swimming, horseback riding, fitness training, and ball games.

Instrumental activities of daily living (IADL) are the skills needed to perform tasks associated with an independent lifestyle. Not all teens or adults with AMC reach a level of independence that allows autonomous living. The use of regular or adapted public transport, or the process for obtaining a driver's license, is critical and usually necessary for education and employment roles. The limitations of ROM experienced by persons with AMC can make household tasks such as cooking, laundry, and cleaning very challenging. Steen et al. (2017) correlated physical active and passive ROM of upper and lower extremity joints. They found that shoulder active and passive motion correlated with eating and drinking. Shoulder active ROM plus wrist active and passive supination was helpful for dressing. Dressing and bathing often required assistance, but individuals were creative in finding ways to do many IADLs. Adults with AMC may continue to use durable medical equipment like wheelchairs and bath seats. Access to a specialty clinic, general hospital, or private vendor will help with timely repairs or prescriptions for new equipment, which can prevent injury or accident. Participation in support groups, internet searches, and healthcare collaborations can all lead to innovative technical aids and creative solutions to everyday skills.

Rehabilitation in general and therapy goals specifically seek to enhance quality of life (Colver, 2009). Teens and adults should focus on reaching education/vocational/leisure goals. The HCP can assist through adaptations and problem-solving strategies as well as through facilitating employment and managing workplace environments.

6 | OPPORTUNITIES FOR GROWTH

The rehabilitation journey for AMC begins in the infant stages and continues throughout the lifespan. There is a constant need to address the mobility of the stiff joints; but, the approach varies throughout the developmental process. Opportunities for growth include exploring alternative treatment techniques. Nontraditional therapies such as craniosacral therapy and myofascial therapy have limited objective research to support their use; however, subjective reports describe improvement when used. Other options that have evidence-based support for use in children, but not specific to AMC, include treadmill training, aquatic therapy, and neuromuscular electrical stimulation. Little information is available regarding the frequency and duration of specific rehabilitation exercises for children with AMC. Single reports suggest that early intensive rehabilitation, supported by daily supervised exercises may give the child an opportunity to improve ROM in the joints and reduce the need for surgical intervention (Ayadi et al., 2015; Azbell & Dannemiller, 2015; Binkiewicz-Glinska et al., 2016; Sells et al., 1996). While ROM is critical, it is equally important for children with AMC to participate in play and leisure activities with their peers. The balance of encouraging play and independence with formal therapy and exercise is important. Formal collaboration among team members on exercise programs, adaptations, or potential surgical interventions is lacking in most healthcare settings. Development of standards of care or a framework for the management of children with AMC would assist with consistent care among all providers.

Along with the challenge to develop standards of care, there is a need to develop and psychometrically test evaluation tools specific to the needs of the AMC population. Implementation of valid, reliable, diagnosis-specific outcome measures would allow the ability to formulate objective conclusions and compare various therapeutic approaches.

Review of the literature reveals poor levels of evidence and limited data due to the rare, and heterogeneous presentation of this population (Miller & Sawatzky, 2017). Opportunities for growth should focus on multi-center site partnerships to evaluate these outcomes and standards of care. Although each of the persons living with AMC is different and the paths taken throughout the journey are varied, the common destination of rehabilitation is maximum independence and participation in life.

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REFERENCES

Abdel-Ghani, H., Mahmoud, M., Shaheen, A., & Abdel-Wahed, M. (2017). Treatment of congenital clasped thumb in arthrogryposis. *The Journal of Hand Surgery, European Volume*, *42*, 794–798. https://doi.org/10.1177/1753193417712863

Alexander, R., Boehme, R., & Cupps, B. (1993). *Normal development of functional motor skills: The first year of life*. (n.p.). Published by Communication Skill Builders/Therapy Skill Builders: Hammill Institute on Disabilities.

Ayadi, K., Trigui, M., Abid, A., Cheniour, A., Zribi, M., & Keskes, H. (2015). Arthrogryposis: Clinical manifestations and management. *Archives de Pédiatrie*, *22*, 830–839. https://doi.org/10.1016/j.arcped.2015.05.014

Azbell, K., & Dannemiller, L. (2015). A case report of an infant with arthrogryposis. *Pediatric Physical Therapy*, *27*, 293–301. https://doi.org/10.1097/PEP.0000000000000148

Babik, I., Kokkoni, E., Cunha, A. B., Galloway, J. C., Rahman, T., & Lobo, M. A. (2016). Feasibility and effectiveness of a novel exoskeleton for an infant with arm movement impairments. *Pediatric Physical Therapy*, *28*, 338–346. https://doi.org/10.1097/PEP.0000000000000271

Banas, B. B., & Gorgon, E. J. (2014). Clinimetric properties of sitting balance measures for children with cerebral palsy: A systematic review. *Physical & Occupational Therapy in Pediatrics*, *34*, 313–334. https://doi.org/10.3109/01942638.2014.881952

Bartonek, A. (2015). The use of orthoses and gait analysis in children with AMC. *Journal of Children's Orthopaedics*, *9*, 437–447. https://doi.org/10.1007/s11832-015-0691-7

Bartonek, A., Eriksson, M., & Gutierrez-Farewik, E. M. (2007). Effects of carbon fibre spring orthoses on gait in ambulatory children with motor disorders and platarflexor weakness. *Developmental Medicine and Child Neurology*, *49*, 615–620.

Binkiewicz-Glinska, A., Sobierajska-Rek, A., Bakula, S., Wierzba, J., Drewek, K., Kowalski, I. M., & Zaborowska-Sapeta, K. (2013). Arthrogryposis in infancy, multidisciplinary approach: Case report. *BMC Pediatrics*, *13*, 184. https://doi.org/10.1186/1471-2431-13-184

Binkiewicz-Glinska, A., Wierzba, J., Szurowska, E., Ruckeman-Dziurdzinska, K., Bakula, S., Sokolow, M., & Renska, A. (2016). Arthrogryposis multiplex congenital - multidisciplinary care - including own experience. *Developmental Period Medicine*, *20*, 191–196.

Bohm, H., Dussa, C. U., Multerer, C., & Doderlein, L. (2013). Pathological trunk motion during walking in children with amyoplasia: Is it caused by muscular weakness or joint contractures? *Research in Developmental Disabilities*, *34*, 4286–4292. https://doi.org/10.1016/j.ridd.2013.09.020

Carlson, W. O., Speck, G. J., Vicari, V., & Wenger, D. R. (1985). Arthrogryposis multiplex Congenita: A long-term follow-up study. *Clinical Orthopaedics and Related Research*, *194*, 115–124.

Carson, V., Hunter, S., Kuzik, N., Wiebe, S. A., Spence, J. C., Friedman, A., … Hinkley, T. (2016). Systematic review of physical activity and cognitive development in early childhood. *Journal of Science and Medicine in Sport*, *19*, 573–578. https://doi.org/10.1016/j.jsams.2015.07.011

Centers for Disease Control and Prevention. (2018). Increasing physical activity among adults with disabilities. Retrieved from https://www.cdc.gov/ncbddd/disabilityandhealth/pa.html

Colver, A. (2009). Quality of life and participation. *Developmental Medicine and Child Neurology*, *51*, 656–659. https://doi.org/10.1111/j.1469-8749.2009.03321.x

Dillon, E. R., Bjornson, K. F., Jaffe, K. M., Hall, J. G., & Song, K. (2009). Ambulatory activity in youth with arthrogryposis: A cohort study. *Journal of Pediatric Orthopedics*, *29*, 214–217. https://doi.org/10.1097/BPO.0b013e3181990214

Donohoe, M. (2006). Arthrogryposis multiplex Congenita. In S. K. Campbell, D. W. Wander Linden, & R. J. Palisano (Eds.), *Physical therapy for children* (3rd ed., pp. 381–400). St. Louis: Saunders Elsevier.

Donohoe, M., Pruszcynski, B., Rogers, K., & Bowen, J. R. (2019). Predicting ambulatory function based on infantile lower extremity posture types in Amyoplasia Arthrogryposis. *Journal of Pediatric Orthopaedics Advance Online Publication.*, *39*, e531–e535. https://doi.org/10.1097/BPO.0000000000001322

Eriksson, M., Gutierrez-Farewik, E. M., Brostrom, E., & Bartonek, A. (2010). Gait in children with Arthrogryposis multiplex Congenita. *Journal of Children's Orthopaedics*, *4*(1), 21–31. https://doi.org/10.1007/s11832-009-0234-1

Eriksson, M., Jylli, L., Villard, L., Kroksmark, A. K., & Bartonek, A. (2018). Health-related quality of life and orthosis use in a Swedish population with Arthrogryposis. *Prosthetics and Orthotics International*, *42*(4), 402–409. https://doi.org/10.1177/0309364618774059

Gmelig Meyling, C., Ketelaar, M., Kuijper, M. A., Voorman, J., & Buizer, A. I. (2018). Effects of postural management on hip migration in children with cerebral palsy: A systematic review. *Pediatric Physical Therapy*, *30*(2), 82–91. https://doi.org/10.1097/PEP.0000000000000488

Graubert, C. S., Chaplin, D. L., & Jaffe, K. M. (1998). Physical and occupational therapy. In L. T. Staheli, J. G. Hall, K. M. Jaffe, & D. O. Paholke (Eds.), *Arthrogryposis: A text atlas* (pp. 87–113). Cambridge: Cambridge University Press.

Hall, J. G. (2014). Arthrogryposis (multiple congenital contractures): Diagnostic approach to etiology, classification, genetics, and general principles. *European Journal of Medical Genetics*, *57*(8), 464–472. https://doi.org/10.1016/j.ejmg.2014.03.008

Joubert, F., & Franzsen, D. (2015). Self-care of children with Arthrogryposis in Gauteng, South Africa. *British Journal of Occupational Therapy*, *79*(1), 35–41. https://doi.org/10.1177/0308022615580327

Kaplan, S. L., Coulter, C., & Fetters, L. (2013). Physical therapy management of congenital muscular torticollis: An evidence-based clinical practice guideline: From the section on Pediatrics of the American Physical Therapy Association. *Pediatric Physical Therapy*, *25*(4), 348–394 doi: 10.1097PEP.0b013e3182a778d2.

Kowalczyk, B., & Felus, J. (2016). Arthrogryposis: An update on clinical aspects, etiology, and treatment strategies. *Archives of Medical Science*, *12*(1), 10–24. https://doi.org/10.5114/aoms.2016.57578

Kroksmark, A. K., Kimber, E., Jerre, R., Beckung, E., & Tulinius, M. (2006). Muscle involvement and motor function in amyoplasia. *American Journal of Medical Genetics. Part A*, *140*(16), 1757–1767. https://doi.org/10.1002/ajmg.a.31387

Lake, A. L., & Oishi, S. N. (2015). Hand therapy following elbow release for passive elbow flexion and long head of the triceps transfer for active elbow flexion in children with amyoplasia. *Journal of Hand Therapy*, *28*(2), 222–226; quiz 227. https://doi.org/10.1016/j.jht.2014.10.007

Martin Ginis, K. A., van der Scheer, J. W., Latimer-Cheung, A. E., Barrow, A., Bourne, C., Carruthers, P., … Goosey-Tolfrey, V. L. (2018). Evidence-based scientific exercise guidelines for adults with spinal cord injury: An update and a new guideline. *Spinal Cord*, *56*(4), 308–321. https://doi.org/10.1038/s41393-017-0017-3

McKeever, P., Rossen, B. E., Scott, H., Robinson-Vincent, K., & Wright, V. (2013). The significance of uprightness: parents' reflections on children's responses to a hands-free walker for children. *Disability & Society*, *28*(3), 380–392. https://doi.org/10.1080/09687599.2012.714259

Miller, R., & Sawatzky, B. (2017). Outcomes at 2-year minimum follow up of shoulder, elbow and wrist surgery in individuals with arthrogryposis multiplex congenita. *Journal of Clinical & Experimental Orthopaedics*, *3*(1). https://doi.org/10.4172/2471-8416.100028

Nouraei, H., Sawatzky, B., MacGillivray, M., & Hall, J. (2017). Long-term functional and mobility outcomes for individuals with Arthrogryposis multiplex Congenita. *American Journal of Medical Genetics. Part A*, *173*(5), 1270–1278. https://doi.org/10.1002/ajmg.a.38169

Paleg, G. S., Smith, B. A., & Glickman, L. B. (2013). Systematic review and evidence-based clinical recommendations for dosing of pediatric supported standing programs. *Pediatric Physical Therapy*, *25*(3), 232–247. https://doi.org/10.1097/PEP.0b013e318299d5e7

Palmer, P. M., MacEwen, G. D., Bowen, J. R., & Mathews, P. A. (1985). Passive motion therapy for infants with arthrogryposis. *Clinical Orthopaedics and Related Research*, *194*, 54–59.

Park, E. S., Park, C. I., & Kim, J. Y. (2001). Comparison of anterior and posterior walkers with respect to gait parameters and energy expenditure of children with spastic diplegic cerebral palsy. *Yonsei Medical Journal*, *42*(2), 180–184.

Sells, J. M., Jaffe, K. M., & Hall, J. G. (1996). Amyoplasia, the most common type of Arthrogryposis: The potential for good outcome. *Pediatrics*, *97*(2), 225–231.

Sival, D. A., Prechtl, H. F. R., Sonder, G. H. A., & Touwen, B. C. L. (1993). The effect of intra-uterine breech position on postnatal motor functions of the lower limbs. *Early Human Development*, *32*(2–3), 161–176.

Staheli, L. T., Hall, J. G., Jaffe, K. M., & Paholke, D. O. (1998). *Arthrogryposis: A text atlas*. Cambridge: Cambridge University Press.

Steen, U., Christensen, E., & Samargian, A. (2017). Adults living with Amyoplasia: Function, psychosocial aspects, and the benefit of AMC support groups. *The Journal of Pediatric Orthopedics*, *37*(5), S31–S32. https://doi.org/10.1097/BPO.0000000000001003

Stief, F., Bohm, H., Ebert, C., Doderlein, L., & Meurer, A. (2014). Effect of compensatory trunk movements on knee and hip joint loading during gait in children with different orthopedic pathologies. *Gait & Posture*, *39*(3), 859–864. https://doi.org/10.1016/j.gaitpost.2013.11.012

van Bosse, H. J. P., Ponten, E., Wada, A., Agranovich, O. E., Kowalczyk, B., Lebel, E., … Durgut, F. (2017). Treatment of the lower extremity contracture/deformities. *Journal of Pediatric Orthopedics*, *37*(Suppl 1), S16–S23. https://doi.org/10.1097/BPO.0000000000001005

Van Heest, A., James, M. A., Lewica, A., & Anderson, K. A. (2008). Posterior elbow capsulotomy with triceps lengthening for treatment of elbow extension contracture in children with Arthrogryposis. *The Journal of Bone and Joint Surgery. American Volume*, *90*(7), 1517–1523. https://doi.org/10.2106/JBJS.F.01174

Vermaak, D. P. (2012). Arthrogryposis multiplex Congenita of the upper limb. *SA Orthopaedic Journal*, *11*(1), 34–39.

World Health Organization. (2001). *International Classificaton of functioning, disability, and health*. Geneva, Switzerland: World Health Organization. Retrieved from. https://www.who.int/classifications/icf/en/

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