Received: 25 June 2019 | Revised: 31 July 2019 | Accepted: 1 August 2019

DOI: 10.1002/ajmg.c.31734

RESEARCH ARTICLE

AMERICAN JOURNAL OF medical genetics PART C Seminars in Medical Genetics

WILEY

Treatment and outcomes of arthrogryposis in the lower extremity

Reggie C. Hamdy[1,2] | Harold van Bosse[3] | Haluk Altiok[4] | Khaled Abu-Dalu[5] | Pavel Kotlarsky[5] | Alicja Fafara[6,7] | Mark Eidelman[5]

[1]Shriners Hospitals for Children, Montreal, Québec, Canada

[2]Department of Pediatric Orthopaedic Surgery, Faculty of Medicine, McGill University, Montreal, Québec, Canada

[3]Shriners Hospitals for Children, Philadelphia, Pennsylvania

[4]Shriners Hospitals for Children, Chicago, Illinois

[5]Pediatric Orthopedics, Technion Faculty of Medicine, Ruth Children's Hospital, Haifa, Israel

[6]Faculty of Health Science, Institute of Physiotherapy, Jagiellonian University Medical College, Krakow, Poland

[7]Arthrogryposis Treatment Centre, University Children's Hospital, Krakow, Poland

Correspondence
Reggie C. Hamdy, Shriners Hospitals for Children, Montreal, Québec, Canada.
Email: rhamdy@shriners.mcgill.ca

Abstract

In this multiauthored article, the management of lower limb deformities in children with arthrogryposis (specifically Amyoplasia) is discussed. Separate sections address various hip, knee, foot, and ankle issues as well as orthotic treatment and functional outcomes. The importance of very early and aggressive management of these deformities in the form of intensive physiotherapy (with its various modalities) and bracing is emphasized. Surgical techniques commonly used in the management of these conditions are outlined. The central role of a multidisciplinary approach involving all stakeholders, especially the families, is also discussed. Furthermore, the key role of functional outcome tools, specifically patient reported outcomes, in the continuous monitoring and evaluation of these deformities is addressed. Children with arthrogryposis present multiple problems that necessitate a multidisciplinary approach. Specific guidelines are necessary in order to inform patients, families, and health care givers on the best approach to address these complex conditions

KEYWORDS

lower limb, surgery, rehabilitation, orthotics, functional outcomes

1 | INTRODUCTION

The overall goal in the management of patients with arthrogryposis is optimization of quality of life by rendering these patients as independent as possible in their activities of daily living and by attaining independent ambulation and ultimately independent living (Kowalczyk & Felus, 2015). In order to attain this goal, a comprehensive, multidisciplinary, very early, and aggressive approach is warranted.

This comprehensive approach is complex, necessitates the expertise of numerous medical and para-medical personnel, and hence, should be embedded within a multidisciplinary program. The family and caretakers should be at the center of this approach. It cannot be over emphasized that treatment should be very aggressive and should start immediately after birth. This includes three components: rehabilitation, orthotics, and surgical approaches. A general plan of treatment should be developed and communicated to the family, in order for them to actively participate in the treatment and thus offer the best chances of success.

Unlike many other neuromuscular conditions, patients with arthrogryposis have normal sensation and IQ, and the functional long-term prognosis is usually good. The contractures are usually most severe at birth and, with early, aggressive adequate treatment, they gradually improve with life. This has to be clearly explained to the family. It has been shown that long-term functional outcome and ambulatory status at skeletal maturity and in adulthood are not necessarily correlated with the severity of the condition at birth and because of this, early aggressive treatment is warranted (Altiok, Flanagan, Krzak, & Hassani, 2019; Fassier, Wicart, Dubousset, & Seringe, 2009; Nouraei, Sawatzky, MacGillivray, & Hall, 2017).

In this multiauthored article, deformities of the lower limb as it pertains to patients with Amyoplasia (the most common form of arthrogryposis) will be discussed. Although lower limb function is

important for independent ambulation, it is important to emphasize here that it is upper—and not lower limb function—that is the most important determinant of quality of life and independent living. As previously mentioned, upper limb function is crucial for daily living activities such as dressing, perineal hygiene, gripping, and reaching for the mouth for feeding.

In cases of Amyoplasia, the lower limbs are affected in 89 to 95% of cases (Hall, 1997; Hall, Aldinger, & Tanaka, 2014; Hall, Reed, & Driscoll, 1983; Sells, Jaffe, & Hall, 1996). The foot is most commonly affected in about 90% of cases, followed by knee deformities in about 70% of cases and then hip deformities in about 40% of patients (Hamdy & Dahan-Oliel, 2016). All four limbs are usually affected and the degree of deformities and contractures is usually symmetrical. Muscular atrophy and weakness is a hallmark of this condition. Often there is loss of skin creases across the joints and dimpling at the sites of the joints.

A general plan of management should be put forward and this should actively involve the family and all stakeholders. As previously mentioned, this plan should include all three pillars of management of children with arthrogryposis: intensive and aggressive physiotherapy, bracing, and surgical interventions. It is often recommended that the foot deformities be treated first, followed by the knee and finally the hip (Bernstein, 2002; Hamdy & Dahan-Oliel, 2016). Therefore, this article presents management of the ankle and feet, knees and hips, as well as considerations for bracing, physical therapy, and measuring function of the lower extremities in arthrogryposis.

2 | ANKLE AND FOOT DEFORMITIES IN ARTHROGRYPOSIS

Deformities of the foot are common in all types of arthrogryposis. The most common deformity is severe clubfoot, with a reported incidence of up to 90% (Figure 1). Other deformities include congenital vertical talus (CVT; 3–10%) and, less often, isolated equinus, cavovarus, equinovalgus, metatarsus adductus, and calcaneovalgus (Bevan et al., 2007; Guidera & Drennan, 1985; Sodergard & Ryoppy, 1994; van Bosse et al., 2017). The clubfoot deformity can be divided to "classic variant" and equinocavus variant. In equinocavus variant, equinus and midfoot cavus are the main deformities, whereas heel varus, supination of the forefoot, and adductus are very mild (this deformity looks like "atypical" clubfoot; van Bosse, 2015; van Bosse et al., 2017). The difference between syndromic and similar idiopathic deformities is probably the time they formed while in utero (van Bosse, 2015). Deformities in patients with arthrogryposis are characterized by stiffness, resistance to treatment, and a high rate of recurrence (Drummond & Cruess, 1978; Hall et al., 1983; Zimbler & Craig, 1983). Early treatment has been recommended as the soft tissues in the newborn are much suppler and more susceptible to manipulation (van Bosse, 2015). The rate of recurrence following treatment becomes less frequent as the child ages (van Bosse et al., 2017).

The goal of treatment of arthrogrypotic feet is conversion of the deformed foot into a plantigrade and painless platform that can be shoe-able and plantigrade (Lloyd-Roberts & Lettin, 1970). Treatment should be started as soon as possible to allow independent ambulation at an earlier age (Lloyd-Roberts & Lettin, 1970; Niki, Staheli, & Mosca, 1997; Song, 2017). Minimizing the number of surgeries also minimizes the number of recovery periods, providing less disruption to the child's development (Song, 2017; van Bosse, Marangoz, Lehman, & Sala, 2009). Mild residual deformity is also acceptable, as long as the child is pain-free, able to wear regular shoes, and is satisfied with the result (Cassis & Capdevila, 2000). Evaluation of the severity of the clubfoot deformity can be done using the systems of Dimeglio and colleagues (Dimeglio, Bensahel, Souchet, Mazeau, & Bonnet, 1995) or Pirani and colleagues (Pirani, Outerbridge, Sawatzky, & Stothers, 1999). Using these systems allows proper monitoring of the correction process (Kowalczyk & Felus, 2015).

The management of foot deformities in arthrogryposis is difficult because these deformities usually are only part of the problem, as there are also other joint deformities, contractures, and dislocations. Historically, the main treatment for clubfoot in arthrogryposis was surgical. Different surgical treatments were proposed, including soft tissue procedures, with the most popular being posterior and posteromedial release, and bony procedures including talectomies, osteotomies, decancellation of tarsal bones, and fusions. The bony procedures were described both as a primary treatment as well as for revision surgeries. However, the long-term results were unsatisfactory with a high number of recurrences and poor functional outcomes (Carlson, Speck, Vicari, & Wenger, 1985; Chang & Huang, 1997; Drummond & Cruess, 1978; Green, Fixsen, & Lloyd-Roberts, 1984;

FIGURE 1 La pied bot (the clubfoot) by Jose Ribera (1642). This was probably the first depiction of clubfoot in arthrogryposis: note the position of the elbow and typical rigid right clubfoot

Hsu, Jaffray, & Leong, 1984; Menelaus, 1971; Niki et al., 1997; van Bosse, 2015; Widmann, Do, & Burke, 2005; Zimbler & Craig, 1983).

The Ponseti method completely changed the management of idiopathic clubfoot. Several publications showed that the Ponseti method is also effective for treatment of clubfoot in arthrogryposis (Boehm, Limpaphayom, Alaee, Sinclair, & Dobbs, 2008; Janicki et al., 2009; Kowalczyk & Lejman, 2008; Morcuende, Dobbs, & Frick, 2008; Moroney, Noel, Fogarty, & Kelly, 2012; van Bosse, 2015; van Bosse et al., 2009; van Bosse et al., 2017).

Morcuende et al. (2008) described the results of Ponseti treatment in 16 patients (32 feet) with a mean follow-up of more than 4 years. The mean number of casts before Achilles tenotomy was seven, mean amount of post-tenotomy dorsiflexion was 5°. Only one patient did not achieve the treatment goal and underwent posteromedial release. The authors advocate correction of foot abduction to 40–50°, rather than 70° as in the idiopathic clubfoot, in order to reduce stress on the foot and eliminate overcorrection. Despite a relatively small amount of foot dorsiflexion (5° in contrast to 20° for idiopathic clubfoot), many feet improved during follow-up under treatment with a foot abduction brace (FAB; some showed a dramatic improvement at age 3–4 years). During follow-up, four patients underwent soft tissue release. A quarter of patients in the cohort had relapse of the deformity that was not always related to noncompliance with brace use, but due to stiffness stemming from arthrogryposis. This is in contrast to the idiopathic clubfoot where recurrence usually is related to noncompliance with the brace. The authors' conclusion was that, although challenging, the Ponseti method is effective as initial treatment of clubfoot in patients with arthrogryposis.

Boehm and colleagues (Boehm et al., 2008) summarized treatment, using the Ponseti method, of 12 patients (24 feet) with distal arthrogryposis. Initial correction was achieved in all but one patient (92%). Three patients (6 feet) had relapses at an average of 6 month after initial correction. Relapses were defined as more than 5° of hindfoot varus and less than 10° of ankle dorsiflexion. All relapses were related to noncompliance with the use of a FAB. All relapsed patients underwent repeated casting and one underwent repeated Achilles tenotomy.

Van Bosse and colleagues (van Bosse et al., 2009) reviewed the results of treatment, using the Ponseti method, of 10 patients (19 feet). Eight feet had relapse after an average of 38 months. Relapses were treated by repeat casting. Only two patients underwent soft tissue release at latest follow-up.

2.1 | Author's protocol

Treatment of patients with arthrogryposis is always challenging. Rarely are feet and ankle deformities isolated problems, more commonly there is also hip and knee pathology that require treatment.

We always start treatment with modified Ponseti casting, that is, weekly Ponseti above-knee casts applied with 90° of knee flexion. In our opinion, the casting goal should provide a thigh–foot angle of no more than 50°, in contrast to 70° as in idiopathic clubfoot. When the thigh–foot angle is corrected to a desired angle, the foot is ready for Achilles tenotomy. The equinus in arthrogrypotic feet is usually more rigid than in idiopathic congenital talipes equinovarus (CTEV). Therefore, the goal of Achilles tenotomy is to achieve 5–10° of dorsiflexion (in contrast to 20° in idiopathic CTEV). Many children with arthrogryposis start correction of their clubfeet later than their idiopathic counterparts; therefore, the correction of deformities requires more casts. Osteopenia due to prolonged immobilization may cause iatrogenic fractures during manipulations.

FAB in an integral part of the Ponseti method and is especially important in arthrogryposis. During the first 3 months, the patients must wear the brace for approximately 23 hr daily. After this period, the brace should be used for 16 hr daily, until the end of first year.

Relapses are relatively common; therefore, close monitoring is important. In case of a relapse, repeated casting usually solves the problem. Repeated percutaneous Achilles tenotomy might be performed if dorsiflexion is less than 5°.

A combination of rigid hip dislocation with clubfeet is common. In our opinion, clubfeet should be corrected first, followed by open reduction of the hip via the medial approach, combined with Achilles tenotomy (Figures 2–3). The foot abduction brace should be used starting 3 weeks after surgery (Figure 4).

FIGURE 2 (a, b) Six-week-old boy with bilateral CTEV and right hip dislocation. CTEV, congenital talipes equinovarus

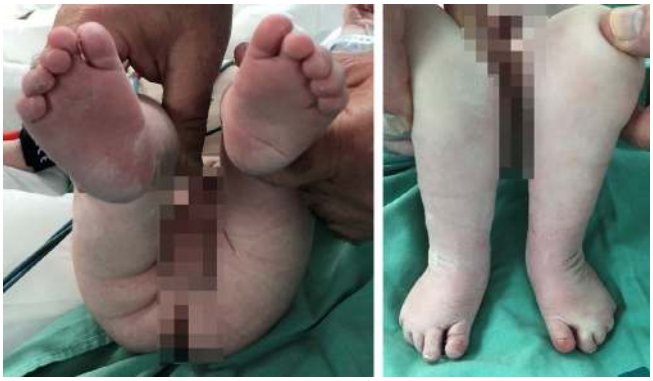

FIGURE 3 Feet after 7 weekly casts: Note that TFA corrected to 50°. TFA, thigh foot angle

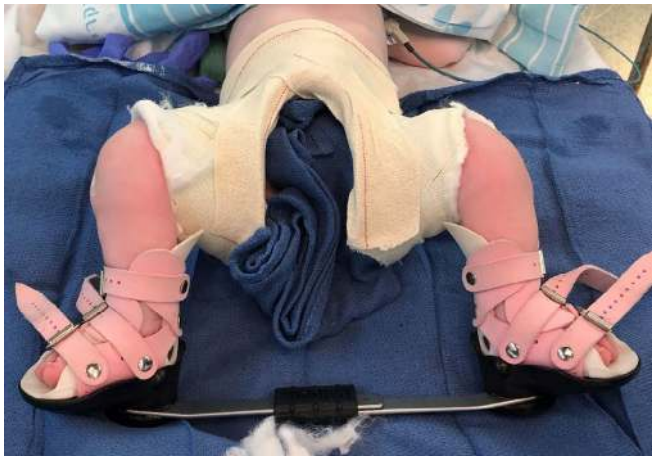

FIGURE 4 Three weeks after open reduction of the right hip via medial approach: Application of foot abduction brace after removal of leg portion of the casts

In rare refractory cases when conservative treatment fails, soft tissue release should be considered. The Cincinnati circular approach around the foot gives the best access to posterior, medial, and lateral structures of the foot and ankle. Saying that, we usually prefer performing a limited posteromedial release as was described by Mosca (Mosca, 2001). We use this operation in feet that are too severe or too late for the regular Ponseti protocol. Limited posteromedial release includes tibialis posterior tendinotomy through a small incision over the midportion of the distal medial leg, percutaneous Achilles tenotomy, percutaneous tenotomy of the flexor hallucis tendon, and, if toe flexors are tight, all flexors divided percutaneously. After that, a long leg cast is applied for 3 weeks followed by treatment using a FAB/ankle–foot orthosis (AFO).

In older children with severe, already operated on or neglected feet, we use circular external fixation. The Ilizarov frame is a classic option; however, we prefer using hexapods. Although all hexapod frames can correct six axis deformities simultaneously using virtual hinges, our preferred option is the Taylor Spatial Frame (Eidelman & Katzman, 2011; Eidelman, Keren, & Katzman, 2012). Supramalleolar osteotomy and posterior angulation might also be an option in severe neglected equinus (Figures 5–10).

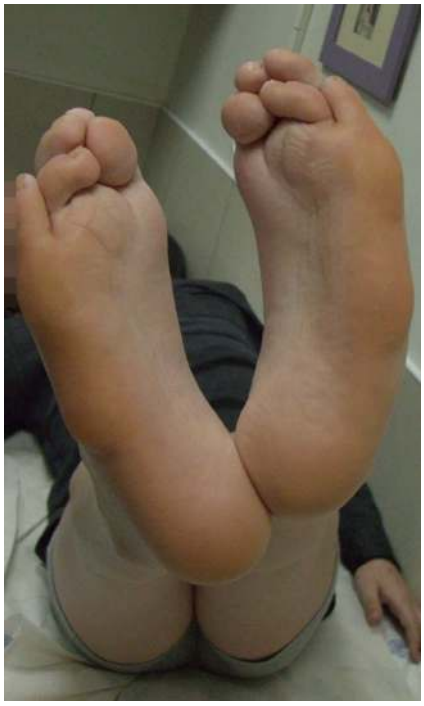

FIGURE 5 Bilateral severe clubfeet

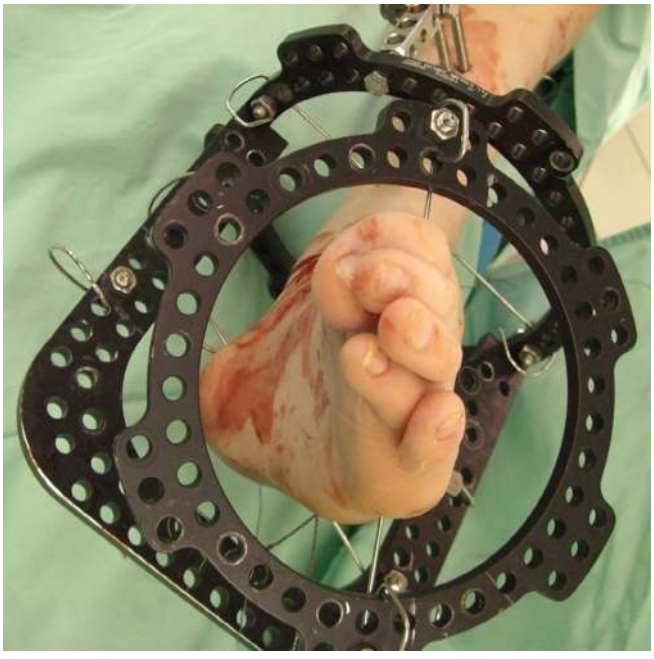

FIGURE 6 Application of Butt frame and midfoot Gigli saw osteotomy

For the treatment of CVT, extensive soft tissue release was the gold standard treatment for many years. The Dobbs method of correction (reversed Ponseti method) changed our approach to the correction of CVT. Today we start treatment with casting as described by Dobbs and colleagues (Chalayon, Adams, & Dobbs, 2012; Dobbs, Purcell, Nunley, & Morcuende, 2006). After several weeks and a weekly change of casts, talonavicular pinning should be performed through a

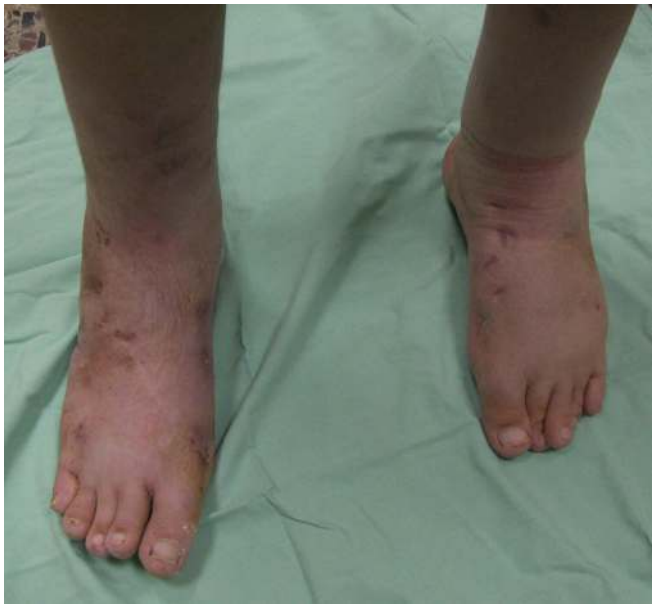

FIGURE 7 After removal of Butt frames

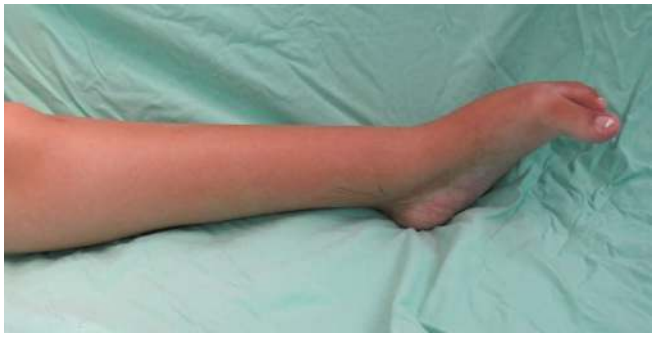

FIGURE 8 Severe bilateral equinus deformity

small medial approach. Postoperatively, molded AFO should be used for several years to prevent relapse.

Even in severe deformities, a majority of feet in arthrogryposis might be treated successfully without extensive soft tissue releases. Monitoring until maturity is imperative to prevent recurrence.

3 | THE KNEE IN CHILDREN WITH ARTHROGRYPOSIS

The knee is the second most commonly affected joint of the lower extremity in children with arthrogryposis (Lampasi, Antonioli, & Donzelli, 2012). Knee flexion contractures are the most commonly encountered deformities, followed by knee extension contractures. Combined flexion and extension contractures can also occur. Physical impairment due to these contractures may be significant. Knee flexion contractures of more than 15–20° may interfere with ambulation and extension contractures may interfere with sitting position (Ponten, 2015).

The goal in the management of these deformities is to reduce the contractures and promote ambulation. An extensive discussion with the family

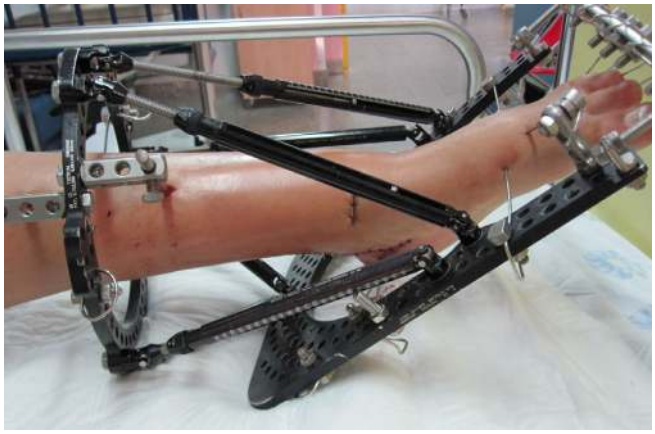

FIGURE 9 Application of TSF and tarsal tunnel release. TSF, Taylor Spatial Frame

is extremely important and it should be clearly explained that these deformities are usually most severe at birth and that—with proper, aggressive and early treatment is warranted, as these contractures will most likely improve with time. However, the recurrence rate of these deformities is usually very high (Yang, Dahan-Oliel, Montpetit, & Hamdy, 2010).

Knee flexion contractures of more than 15° may cause significant functional disabilities (Ponten, 2015), as they may interfere with ambulation and sitting, whereas knee extension deformities are usually better tolerated and may not interfere with walking. It has been shown that correction of knee flexion deformities has a positive effect on the ability to ambulate (Hamdy & Dahan-Oliel, 2016). However, if quadriceps weakness is present and severe, which is often the case in many of children with arthrogryposis, then the gain in the ability to ambulate is minimal, despite correction of the knee flexion deformity.

The management of knee flexion deformities should start immediately after birth in the form of stretching exercises and bracing. If the deformity exceeds 20 to 30° and fails to respond to intense physiotherapy program, then serial corrective casting should be considered. In moderate deformities of 20–40°, soft tissue releases of the medial and lateral hamstrings may not completely correct the deformity and a posterior capsulotomy may be necessary. Recently, anterior hemiepiphysiodesis of the distal femoral growth plate with eight plates has been shown to be a reliable technique in correction of mild and moderate deformities (Figure 11). In severe deformities of more than 40°, supracondylar osteotomy with femoral shortening is an excellent option, as it allows correction of the deformity without putting too much tension on the posterior neurovascular structures. This technique also avoids dissection of the soft tissues at the posterior aspect of the knee. However, there are also disadvantages of this technique. First, it does not increase the range of motion (ROM) of the knee; it only displaces the present arc of motion. Thus, a gain in extension of the knee will also lead to an equivalent decrease in flexion and this may lead to difficulty in the sitting position. Furthermore, it has been shown that there is high recurrence rate of the flexion deformity following supracondylar osteotomies due to rapid remodeling of about 1° per month at the osteotomy site (Figure 12a–d). Therefore, it is

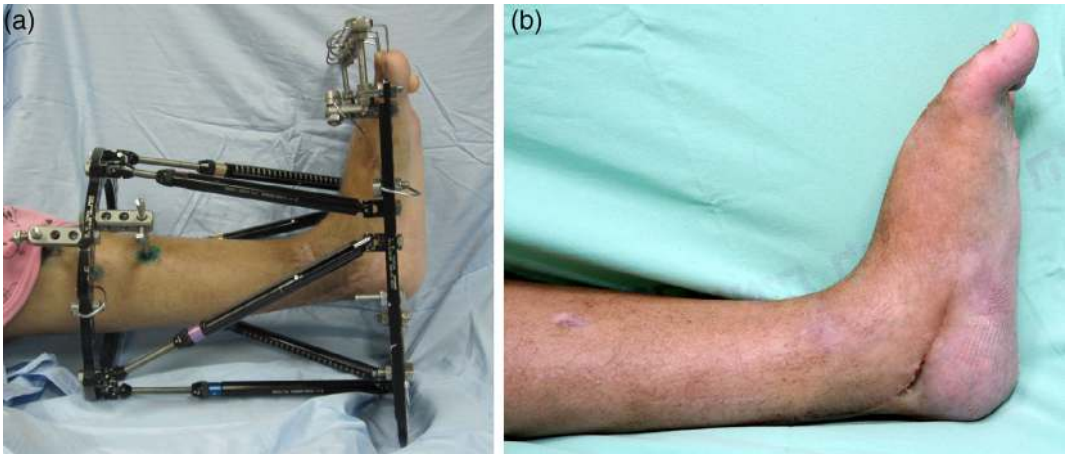

FIGURE 10 (a) At the end of correction, and (b) after removal of the frame

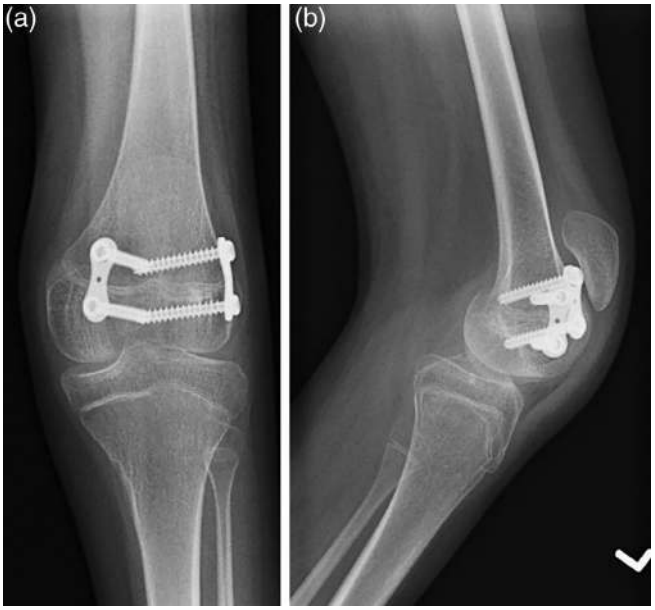

FIGURE 11 Anteroposterior and lateral X-rays of the knee showing anterior hemiepiphysiodesis

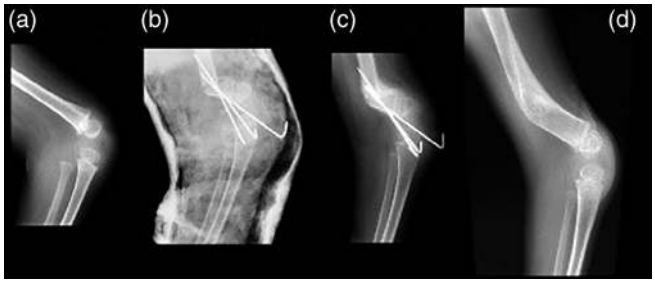

FIGURE 12 (a–d) Supracondylar osteotomy of the femur with satisfactory correction of knee flexion deformity and subsequent remodeling

preferable to perform this osteotomy in patients who are closer to skeletal maturity. Extensive femoral shortening alone without angular correction of the femur is another option. In severe deformities of 60°

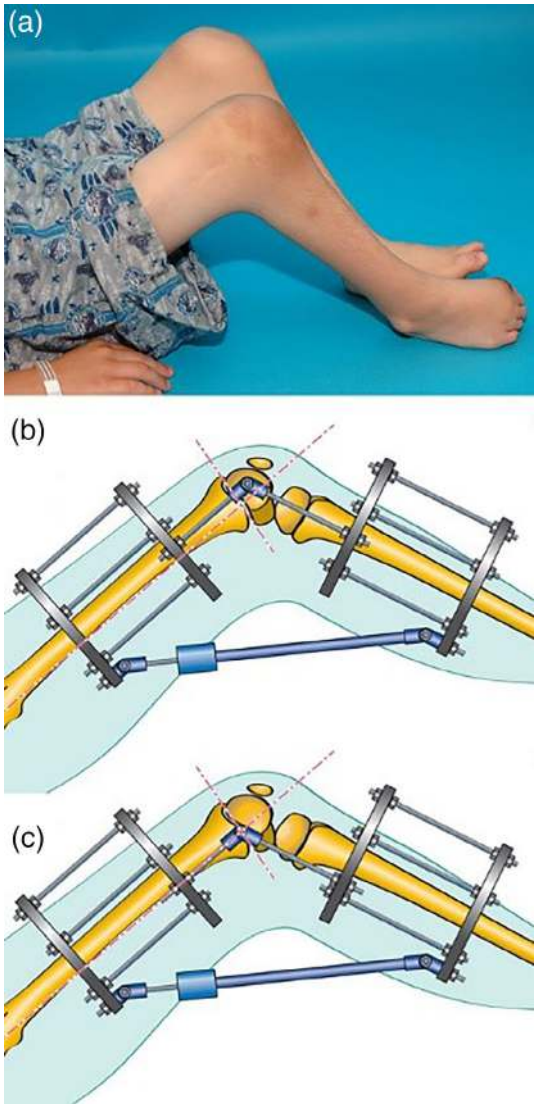

FIGURE 13 (a–c) Circular Ilizarov construct and placement of the hinge for gradual correction of knee flexion deformity

or more and in recurrent deformities, gradual correction of the deformity with external fixators, with or without concomitant posterior soft tissue release, may be preferable to other methods. Contrary to a supracondylar osteotomy, this technique allows an increase in the arc of motion of the knee. Distraction of the knee should be first performed before starting the gradual distraction, in order to avoid compression of the articular cartilage during the correction (Figure 13a–c). Proper preoperative planning and careful attention to surgical details, specifically identification of the center of rotation of the knee and accurate placement of the hinges cannot be overemphasized. This will prevent posterior subluxation of the knee.

Knee extension contractures are less common than flexion deformities and vary in severity from full extension of the knee with various degrees of loss of flexion, hyperextension with recurvatum deformity, knee subluxation, and even frank dislocation. Knee subluxation and dislocation should be treated immediately after birth with passive stretching, corrective casting, and bracing (Figure 14a–c). If this conservative treatment fails then surgical intervention is warranted in the form of percutaneous or mini open tenotomy of the rectus tendon followed by application of a cast with the knee in flexion (Figure 15). If the contracture is not fully corrected, then a release is made medial and lateral to the patella. All para patellar adhesions are released. If still complete correction is not obtained, anterior capsulotomy is performed and if necessary V–Y quadriceps lengthening is performed. In cases of very severe contractures, more extensive release of the extensor mechanism in the form of a Judet quadricepsplasty and femoral shortening could be considered. In cases of knee extension contractures without subluxation or dislocation, there is some controversy as to the ideal time for surgical intervention. Although some authors recommend early intervention during the first year of life, others prefer to wait until the child is older. The rationale for waiting is that knee extension contractures do not interfere with ambulation.

3.1 | Author's protocol

The management of mild knee contracture (0–20°) is physical therapy, stretching, and serial casting. Moderate knee contracture (20–40°) management is anterior distal femoral hemiepiphysiodesis and soft tissue release. Moreover, management of severe knee contractures (40–60°) is femoral extension osteotomy, whereas for very severe knee contractures (60+ degrees), gradual distraction is recommended (Hamdy & Dahan-Oliel, 2016).

4 | HIP DISLOCATIONS AND CONTRACTURES

The hip is a commonly affected joint in children with arthrogryposis, with reported prevalence varying between 30 and 90% of patients (Carlson et al., 1985; Fassier et al., 2009; Friedlander, Westin, & Wood, 1968; Gibson & Urs, 1970; Mead, Lithgow, & Sweeney, 1958; St Clair & Zimbler, 1985; Yau, Chow, Li, & Leong, 2002). Hip contractures without dislocation (18–51%) (Carlson et al., 1985; Fassier et al., 2009; Friedlander et al., 1968; Gibson & Urs, 1970; Lloyd-Roberts & Lettin, 1970; Mead et al., 1958; St Clair & Zimbler, 1985) occur more frequently than congenitally dislocated hips (15–30%; Gibson et al., 1970; Hahn, 1985; Kroksmark, Kimber, Jerre, Beckung, & Tulinius, 2006; Lloyd-Roberts & Lettin, 1970; Mead et al., 1958; St Clair & Zimbler, 1985; Staheli, Chew, & Elliott, 1987; Szoke, Staheli, Jaffe, & Hall, 1996). Correcting these deformities improves the chances of achieving independent ambulatory status, and/or continued functional ambulation as an adult. Hoffer et al. found that hip flexion contractures needed to be less than 30° to allow patients with arthrogryposis functional ambulation (Hoffer, Swank, Eastman, Clark, & Teitge, 1983). The literature is beginning to reflect a more interventionist approach to these hip problems, probably in part due to the recognition of greater ambulatory potential in children with arthrogryposis multiplex congenita (AMC), and observation of continued ambulation late into adulthood.

4.1 | Hip contractures

Hip contractures in arthrogryposis usually include elements of flexion, abduction, and external rotation, although extension contractures do occur. In cases of an isolated mild or moderate hip flexion contracture, a percutaneous release of anterior tethering tissue can be effective (van Bosse et al., 2017). This "percutaneous anterior hip release" is performed through a 10 mm stab incision just medial to the palpable conjoin tendon of the Sartorius and the tensor fascia lata, at the point where it is most palpable inferior to the anterior superior iliac spine. The conjoin tendon is transected by the scalpel blade, and, continuing laterally, the fascia lata is further incised to just superior to the greater trochanter. Residual contractures may require further release of the rectus femoris and/or the fascia overlying the iliopsoas muscle medially if they can be palpated. A Petrie cast is applied, and the remaining

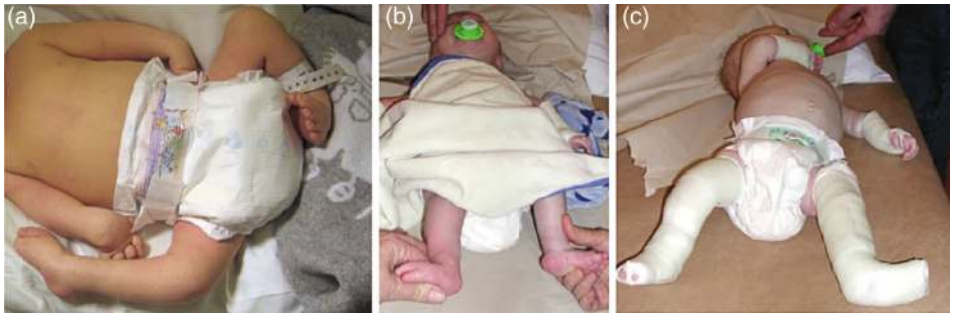

FIGURE 14 (a–c) Newborn with hyperextension deformity and dislocation of the right knee, successfully treated by manipulation and corrective casting

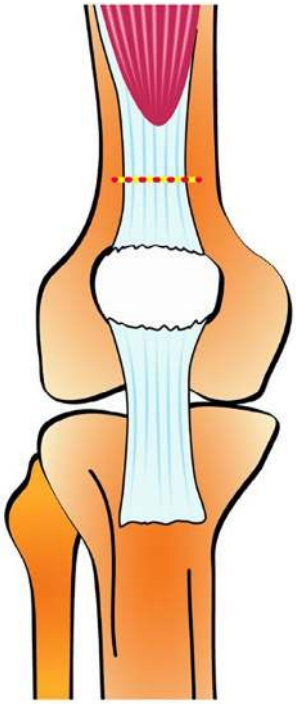

FIGURE 15 Diagram of the knee showing site of transverse cut of the rectus femoris for correction of knee extension deformity

flexion contracture can continue to improve with aggressive physical therapy and prone lying.

When the hip contracture is a multiplanar (flexion, abduction, and external rotation or FAER), typically there is satisfactory overall hip joint motion, but it is poorly positioned for function; a proximal femoral reorientational osteotomy may be needed (van Bosse et al., 2017). The procedure creates a wedge-shaped intratrochanteric osteotomy, aligning the lower extremity with the body, but the hip joint is left in its original position. For those familiar with standard proximal femoral osteotomies, the procedure is relatively straightforward. The limb is positioned in its "natural" or resting position (FAER), and a guide wire is inserted into the femoral neck, parallel to the coronal and transverse planes of the pelvis, regardless of the femoral positioning. Over the guidewire, a cannulated "seating chisel" is passed, with the blade held parallel to the transverse plane of the pelvis. The proximal osteotomy is made just inferior and parallel to the seating chisel, and a second, distal, osteotomy is made perpendicular to the shaft of the femur, removing a wedge of bone. A 90° angled, cannulated blade plate with an offset provides fixation, with the knee rotated directly anteriorly. At an average 40 month follow-up, 36 of 65 patients walked independently, another 20 walked with walkers (van Bosse & Saldana, 2017.

4.2 | Hip dislocations

Hip dislocations in arthrogryposis occur much earlier in gestation than idiopathic hip dislocations and are therefore stiffer with higher displacement. Until recently, the thinking was that reducing arthrogrypotic hips would result in stiff and painful hips, so at most only unilateral hips should be treated to allow for symmetry (Friedlander et al., 1968; Hansen, 1961; Huurman & Jacobsen, 1985; Lloyd-Roberts & Lettin, 1970; Mead et al., 1958; Sarwark, MacEwen, & Scott, 1990; Williams, 1978). But a number of recent studies have good functional success after reduction of congenital dislocations in children with AMC (Bernstein, 2002; Bevan et al., 2007; Staheli et al., 1987; van Bosse et al., 2017; Wada et al., 2012), high rates of independent ambulation (Akazawa et al., 1998; Asif, Umer, Beg, & Umar, 2004; Rocha, Nishimori, Figueiredo, Grimm, & Cunha, 2010; Yau et al., 2002), and good results with both bilateral and unilateral hips (Gruel, Birch, Roach, & Herring, 1986; LeBel & Gallien, 2005; Szoke et al., 1996). A study comparing preoperative and follow-up hip motion in both unilateral and bilateral hip dislocations, the flexion-extension total active motion only decreased by 4° on average. One study of 14 patients with 5–23 years follow-up found 11 were community and 3 were household ambulators. Another study found that 26 of 40 patients ambulated fully independently at an average of 47 months follow-up. Attempted treatment by closed reduction has not been found to be effective in AMC (Robertson, Williamson, & Blattner, 1955; Sarwark et al., 1990; Williams, 1978; Yau et al., 2002).

Different operative strategies championed include performing the reductions early (3–6 months) when a femoral shortening is rarely needed, or reducing late (10 months to 6 years) when a femoral shortening is increasingly necessary, as well as pelvic osteotomies for the older children. Operating later does provide larger structures, which may make a high dislocation easier to correct. The anteriolateral approach has the advantages a direct exposure and accessibility for pelvic osteotomies if needed, but often has higher blood loss and an association with heterotopic bone formation. The medial approach has less blood loss and a more cosmetic scar, but a higher risk of neurovascular traction injury. Stability of the reduced femoral head may be supplemented by using the ligamentum teres to tether of the femoral head to the acetabulum (Wenger, Mubarak, Henderson, & Miyanji, 2008) or by temporarily pinning across the femoral-acetabular joint (Castaneda, Tejerina, Nualart, & Cassis, 2015). Spica cast immobilization for 6 weeks is imperative. Despite radiographic appearances, there is always an appropriately sized acetabulum present. Complications included redislocation and stiffness/lack of hip flexion. Avascular necrosis of Kalamchi and MacEwen Types 2–4 occurred up to 24%, but did not seem to affect hip motion; 80% of avascular necrosis occurred in patients older than 3 years of age suggesting that treatment before 3 years of age is optimal. Future comparison may identify the younger age limit for open reduction.

Although knee flexion contractures occur commonly than extension contractures, the latter are disproportionately associated with dislocated hips among patients with AMC in general. Most patients with dislocated hips have clubfoot deformities, but congenital vertical tali are seen as well. Underlying diagnoses included Amyoplasia, distal arthrogryposis, and central nervous system type of AMC. Bilateral hip dislocations were rarely seen in those with distal arthrogryposis.

4.3 | Author's protocol

In our opinion, management of hip contractures is physical therapy, soft tissue releases and subtrochanteric extension osteotomy (Hamdy & Dahan-Oliel, 2016). However, hip contractures that are abducted, externally rotated, and flexed should be treated with a multiplanar reorientational osteotomy of the proximal femur (van Bosse & Saldana, 2017). Management techniques for hip instability are to reduce unilateral hip dislocation, and in bilateral hip dislocation, surgical treatment depends on mobility of the hip (Hamdy & Dahan-Oliel, 2016).

5 | BRACING THE LOWER EXTREMITY IN ARTHROGRYPOSIS

The role of orthotics to maintain correction and to prevent recurrence is crucial. In individuals with AMC, especially Amyoplasia, the joint contractures can be severe and have a tendency to recur after correction (Vanpaemel, Schoenmakers, van Nesselrooij, Pruijs, & Helders, 1997). Conversely, conservative management and surgical interventions can attempt to address insufficient muscle strength and maintain or augment ROM. A wide spectrum of orthotics can maintain adequate joint position and limb alignment. Orthotic management with corrective splinting during growth has been recommended in the literature to enable independence (Bartonek, 2015; Eriksson, Bartonek, Pontén, & Gutierrez-Farewik, 2015). Each splint/brace is custom-made, so every child or adult with arthrogryposis can function as comfortably as possible and to their fullest potential.

Orthoses are fitted according to each individual's ROM and muscle strength. These orthoses may include AFOs, knee–AFOs (KAFOs), and hip–KAFOs. To enhance or facilitate ambulation in children with AMC, orthoses made of different materials, with or without knee-locking mechanisms (Bartonek, 2015), may be used to compensate for muscle weakness and to support the lower extremities in an aligned position. As such, KAFOs with locked knee joints or AFOs are often used to enable or improve walking function (Bartonek, 2015; Eriksson et al., 2015). Light-weight orthotics are most often needed throughout the adult years, and should be adjusted to address each individual's need for stability, mobility, and power loading (van Bosse et al., 2017). Functional bracing and nighttime splinting are crucial to support ambulation, to compensate for muscle weakness, to keep lower extremities in an aligned position, and to prevent deformity recurrence (Kowalczyk & Feluś, 2016; Lampasi et al., 2012; Ponten, 2015; van Bosse et al., 2017).

6 | PHYSICAL THERAPY

As the World Health Organization's International Classification of Functioning framework (World Health Organization, 2001) indicates, rehabilitation should focus on three levels of treatments: (a) body structure, (b) activity, and (c) participation. Accordingly, physical therapists should work on: (a) optimizing ROM, muscle strength, (b) facilitating self-care and mobility, and (c) enhancing social participation. Physical therapy with children with arthrogryposis should be a balance between those three levels of treatment according to the needs of each neuro-motor developmental stage.

In infants with arthrogryposis, clinical presentation of contractures is the worst at birth. Just after birth, traction and mobilization followed by serial casting could often greatly improve the ROM (Ponten, 2015). Frequent repositioning of an infant helps to improve ROM, encourages the development of head and trunk control, strengthen limb musculatures, and facilitate functional activities. There must be adequate control of the trunk muscles before complex, antigravity movements of the arms and legs are possible (Staheli, Hall, Jaffe, & Paholke, 1998). Throughout the first 3–4 months of life are especially important in activating and stimulating muscle function and in stretching contracted joints to enhance the neuro-development of the baby (Kimber, 2009; Palmer, MacEwen, Bowen, & Mathews, 1985). Daily passive stretching and serial splinting in infants has been found to increase function (Palmer et al., 1985). As the child becomes a toddler, locomotion and other motor abilities develop intensively, ROM and strength of lower limbs muscles must be maintained, and use of equipment (e.g., crutches, canes, and wheelchair) to maximize mobility is explored (Wagner et al., 2019). With increasing age, the frequency of therapy is usually decreased compared to the first year of life (Elfassy et al., 2019), but muscle strengthening and ROM maintenance, as well as functional training, are strongly important.

7 | MEASURING FUNCTION OF THE LOWER EXTREMITY IN ARTHROGRYPOSIS

The heterogeneity inherent to the clinical presentation of arthrogryposis brings challenges for the treating clinician as each individual presents with her or his unique set of musculoskeletal issues, rehabilitation potential, treatment goals, and expected outcomes. The International Classification of Functioning, Disability and Health (ICF) of the World Health Organization provides a theoretical framework and classification system for describing and measuring health and health related states (World Health Organization, 2001). In 2007, the ICF was complemented for children and youth (ICF-CY; World Health Organization, 2007). Like the ICF, the ICF-CY classifies outcome into four domains: body function and structure, activity (i.e., execution of a task or activity), participation (i.e., involvement in a life situation), and environmental and personal factors. When monitoring progress, it is important to distinguish between what a patient is capable of doing in a standardized controlled environment (i.e., capacity) and what the patient actually does in his or her daily environment (i.e., performance; Holsbeeke, Ketelaar, Schoemaker, & Gorter, 2009).

Walking can be examined by both capacity and performance-based measures. Walking capacity is typically measured in the clinic or laboratory and represents what the person is capable of doing in a structured setting. Tools to assess gait capacity and quality include the 6-min walk test, timed up and go test, self-selected 10-min walk test, and three-dimensional (3-D) gait analysis. In contrast, walking performance represents how a person actually performs walking in everyday life. The

Gillette Functional Assessment questionnaire and Functional Mobility Scale are tools that can be used for performance evaluation. Capturing community-based walking (performance) complements the measures that capture the capacity of the individual such as gait analysis and walking tests like 6-min walk test. It could be argued that "what" a child does in their daily life (performance) may be of greater value compared to outcomes measured in the clinic or laboratory (capacity; Ammann-Reiffer, Bastiaenen, de Bie, & van Hedel, 2014; Bjornson, 2019; Himuro, Abe, Nishibu, Seino, & Mori, 2017; Mensch, Rameckers, Echteld, & Evenhuis, 2015; Zanudin, Mercer, Jagadamma, & van der Linden, 2017). To this regard, there has been few studies that used outcome measures to evaluate the capacity and performance of gait in AMC.

Several studies have used outcome measures to describe the activity and participation components of the ICF. Dillon et al. (Dillon, Bjornson, Jaffe, Hall, & Song, 2009) studied a group of children with Amyoplasia and distal arthrogryposis in an attempt to quantify and characterize their activity levels in real-world environment (performance) relative to that of age and sex matched typically developing youth. Participants wore the StepWatch3 Monitor on their ankles for 7 days to monitor the daily frequency, duration, and intensity of their ambulatory activity. The parents of the subjects also completed Activities Scale for Kids, Performance-38 questionnaires to compare parent reported activity levels with StepWatch3 activity. Youth with Amyoplasia and distal arthrogryposis took significantly fewer steps, spent less time at high activity levels, and had significantly lower parental report of ambulatory and physical activity than control group. Eriksson and colleagues (Eriksson, Gutierrez-Farewik, Broström, & Bartonek, 2010) used 3-D gait analysis (capacity) to report on 15 participants who were able to walk, either with or without braces. The 3-D gait analysis showed differences among the participants in trunk, pelvis, and knee kinematics, cadence, and walking speed. In an another study, Eriksson and colleagues (Eriksson, Villard, & Bartonek, 2014) study, energy expenditure and functional exercise capacity were evaluated in 24 children with AMC and 25 typically developing children with the 6-min walk test (6MWT; capacity) and oxygen consumption and cost (efficiency of the gait) using self-selected speed. Participants (mean age = 11.1 years) were independent ambulators with or without orthoses and they all had good muscle strength in their hip extensors and abductors. Only one child had a hip flexion contracture of 20°. Using the FMS, most (n = 19) were classified as community ambulators, and five were classified as household ambulators. The children were divided into three groups based on the type of orthosis they were using. Ten children were using shoes only, whereas the rest had a combination of KAFOs and AFOs. The children walking with open KAFOs or AFOs, most of whom were community ambulators, walked with almost the same walking velocity as AMC children not using orthoses and typically developing children. Their oxygen cost, however, was higher indicating a less efficient gait. The children walking with KAFOs with their knee joint locked had the lowest walking velocity of all groups, and reducing walking velocity in this group was interpreted as a strategy to minimize exertion. During the exercise capacity test in the 6MWT, the children with AMC walked a shorter distance than typically developing children.

Among the many diverse dimensions assessed by the available patient-based outcome measures, physical function has been the main focus in children with Amyoplasia. Amor and colleagues (Amor, Spaeth, Chafey, & Gogola, 2011) studied the utility of Pediatric Outcomes Data Collection Instrument (PODCI) as a tool to evaluate the functional outcomes of children with Amyoplasia. The PODCI contains six domains: upper extremity functioning, transfers and basic mobility, sports and physical functioning, comfort or pain, happiness with physical condition, and global functioning. The questionnaires were administered to parents of patients aged 2 to 18 years. A total of 74 patients with Amyoplasia were included in the study. The mean scores for children with Amyoplasia were significantly lower than the mean scores for children without musculoskeletal disorders in all six domains and was sensitive to changes in function over time. Eriksson and colleauges (Eriksson, Jylli, Villard, Kroksmark, & Bartonek, 2018) evaluated the health-related quality of life in children with AMC and satisfaction with orthoses. Questionnaires were used which measured health-related quality of life using the Child Health Questionnaire-Parent Form (CHQ, EQ-5D-Youth version), mobility and self-care using the Pediatric Evaluation of Disability Inventory (PEDI), and satisfaction with orthoses using the Quebec User Evaluation of Satisfaction with Assistive Technology 2.0. PEDI scores were poorer for those with orthoses than those without. CHQ parent-report scores were significantly lower in 9 of 12 subscales compared with healthy controls. In total, both orthoses groups were quite satisfied with their orthoses. In a study by Ho and Karol (Ho & Karol, 2008), 32 patients with AMC were evaluated to study the utility of knee releases. Patients were assessed using the FMS, PODCI, PEDI, and the Functional Independence Measure for Children (WeeFIM). Whereas knee releases did improve function in the short term, function and outcomes declined as patients aged. Patients with arthrogryposis demonstrated significant impairment in normative scores for upper extremity, physical function, transfers/mobility, sports/physical function, and global function domains. In addition, function as measured by the PODCI, WeeFIM, and PEDI showed decreased scores as length of follow-up increased.

Among adults with AMC, Jones and colleagues (Jones, Miller, Street, & Sawatzky, 2019) demonstrated that the Oswestry Disability Index is a valid outcome tool for low-back and lower extremity pain-related disability. Nouraei et al. (2017), in a study of 177 adults with AMC, demonstrated the utility of two outcome measures: Short Form-36 for quality of life and Physical Activity Scale for Individuals with Physical Disabilities for physical activity. Although the patients reported a quality of life comparable to the general U.S. population, physical function and vitality scores were somewhat less. The average MET score for the level of activity of the study participants were considerably lower than a fit healthy able-bodied individual, 13.5 versus 22 METS, respectively.

Achieving, restoring, or sustaining the ability to walk is one of the main goals for children and adults with Amyoplasia. To evaluate interventions and monitor progress, valid, reliable, and responsive assessment tools are essential. Little information exits that would inform the health care providers and the families what health status changes are of value to the affected individuals. Based on the studies mentioned

here, future research should evaluate the ambulatory function of individuals with Amyoplasia in their social environment. It is increasingly recognized that traditional biomedical outcomes, such as clinical and laboratory measures, need to be complemented by measures that focus on patients' concerns in order to evaluate interventions and provide client-centered care (Fitzpatrick, Davey, Buxton, & Jones, 1998). Patient-reported outcomes are questionnaires or assessment tools that patients complete by themselves or by a family member when self-report is not possible to gain an understanding of their experiences and concerns in relation to health status, health-related quality of life and treatment effectiveness. There is currently no condition-specific outcome measure for individuals with AMC. There are already a number of validated tools to be used among children with cerebral palsy and spinal cord injury, but these tools need to be validated for children with Amyoplasia. Measures that were validated for populations other than AMC (i.e., the Functional Mobility Scale was developed for children with cerebral palsy), should be validated for use in arthrogryposis to ensure proper measurement. Further research is warranted to guide clinicians and researchers regarding the use of outcome measures for children with arthrogryposis.

8 | CONCLUSION

In the management of lower limb deformities in children with arthrogryposis (specifically Amyoplasia), very early and aggressive management of these deformities in the form of intensive physiotherapy and bracing is important. Surgery is performed to improve functional outcomes. Children with arthrogryposis present multiple problems that necessitate a multidisciplinary approach. Specific guidelines are necessary in order to inform patients, families, and health care givers on the best approach to address these complex conditions.

ORCID

Reggie C. Hamdy https://orcid.org/0000-0002-0664-2843
Alicja Fafara https://orcid.org/0000-0003-0641-846X

REFERENCES

Akazawa, H., Oda, K., Mitani, S., Yoshitaka, T., Asaumi, K., & Inoue, H. (1998). Surgical management of hip dislocation in children with arthrogryposis multiplex congenita. *The Journal of Bone and Joint Surgery: The British Volume.*, *80*(4), 636–640.

Altiok, H., Flanagan, A., Krzak, J. J., & Hassani, S. (2019). Quality of life, satisfaction with life, and functional mobility of young adults with arthrogryposis after leaving pediatric care. *American Journal of Medical Genetics*. https://doi.org/10.1002/ajmg.c.31717 [Epub ahead of print].

Ammann-Reiffer, C., Bastiaenen, C. H., de Bie, R. A., & van Hedel, H. J. (2014). Measurement properties of gait-related outcomes in youth with neuromuscular diagnoses: A systematic review. *Physical Therapy*, *94*(8), 1067–1082. https://doi.org/10.2522/ptj.20130299

Amor, C. J., Spaeth, M. C., Chafey, D. H., & Gogola, G. R. (2011). Use of the pediatric outcomes data collection instrument to evaluate functional outcomes in arthrogryposis. *Journal of Pediatric Orthopaedics*, *31*(3), 293–296. https://doi.org/10.1097/BPO.0b013e31820cad93

Asif, S., Umer, M., Beg, R., & Umar, M. (2004). Operative treatment of bilateral hip dislocation in children with arthrogryposis multiplex congenita. *Journal of Orthopaedic Surgery (Hong Kong)*, *12*(1), 4–9.

Bartonek, A. (2015). The use of orthoses and gait analysis in children with AMC. *Journal of Children's Orthopaedics*, *9*(6), 437–447. https://doi.org/10.1007/s11832-015-0691-7

Bernstein, R. M. (2002). Arthrogryposis and amyoplasia. *The Journal of the American Academy of Orthopaedic Surgeons*, *10*(6), 417–424.

Bevan, W. P., Hall, J. G., Bamshad, M., Staheli, L. T., Jaffe, K. M., & Song, K. (2007). Arthrogryposis multiplex congenita (amyoplasia): An orthopaedic perspective. *Journal of Pediatric Orthopaedics*, *27*, 594–600. https://doi.org/10.1097/BPO.0b013e318070cc76

Bjornson, K.F. (2019). Measurement of community-based walking activity in cerebral palsy. *Developmental Medicine and Child Neurology*. 2019 Mar 25. [Epub ahead of print]. doi: https://doi.org/10.1111/dmcn.14226.

Boehm, S., Limpaphayom, N., Alaee, F., Sinclair, M. F., & Dobbs, M. B. (2008). Early results of the Ponseti method for the treatment of clubfoot in distal arthrogryposis. *Journla of Bone and Joint Surgery: American Volume.*, *90*(7), 1501–1507. https://doi.org/10.2106/JBJS.G.00563

Carlson, W. O., Speck, G. J., Vicari, V., & Wenger, D. R. (1985). Arthrogryposis multiplex congenita. A longterm follow-up study. *Clinical Orthopaedics and Related Research*, *194*, 115–123.

Cassis, N., & Capdevila, R. (2000). Talectomy for clubfoot in arthrogryposis. *Journal of Pediatric Orthopaedics*, *20*(5), 652–655.

Castaneda, P., Tejerina, P., Nualart, L., & Cassis, N. (2015). The safety and efficacy of a transarticular pin for maintaining reduction in patients with developmental dislocation of the hip undergoing an open reduction. *Journal of Pediatric Orthopaedics*, *35*(4), 358–362. https://doi.org/10.1097/BPO.0000000000000284

Chalayon, O., Adams, A., & Dobbs, M. B. (2012). Minimally invasive approach for the treatment of non-isolated congenital vertical talus. *The Journal of Bone and Joint Surgery. American Volume*, *94*(11), e73–e77. https://doi.org/10.2106/JBJS.K.00164

Chang, C. H., & Huang, S. C. (1997). Surgical treatment of clubfoot deformity in arthrogryposis multiplex congenita. *Journal of the Formosan Medical Association*, *96*(1), 30–35.

Dillon, E. R., Bjornson, K. F., Jaffe, K. M., Hall, J. G., & Song, K. (2009). Ambulatory activity in youth with arthrogryposis. *Journal of Pediatric Orthopaedics*, *29*(2), 214–217. https://doi.org/10.1097/BPO.0b013e3181990214

Dimeglio, A., Bensahel, H., Souchet, P., Mazeau, P., & Bonnet, F. (1995). Classification of clubfoot. *Journal of Pediatric Orthopaedic Surgery Part B.*, *4*(2), 129–136.

Dobbs, M. B., Purcell, D. B., Nunley, R., & Morcuende, J. A. (2006). Early results of a new method of treatment for idiopathic congenital vertical talus. *Journal of Bone and Joint Surgery. American Volume*, *88*, 1192–1200.

Drummond, D. S., & Cruess, R. L. (1978). The management of the foot and ankle in arthrogryposis multiplex congenita. *Journal of Bone and Joint Surgery. British Volume (London)*, *60*(1), 96–99.

Eidelman, M., & Katzman, A. (2011). Treatment of arthrogrypotic foot deformities with the Taylor Spatial Frame. *Journal of Pediatric Orthopedics*, *31*, 429–434. https://doi.org/10.1097/BPO.0b013e3182172392

Eidelman, M., Keren, Y., & Katzman, A. (2012). Correction of residual clubfoot deformities in older children using the Taylor spatial butt frame and midfoot Gigli saw osteotomy. *Journal of Pediatric Orthopedics*, *32*, 527–533. https://doi.org/10.1097/BPO.0b013e318259ff2d

Elfassy, C., Darsaklis, V. B., Snider, L., Gagnon, C., Hamdy, R., & Dahan-Oliel, N. (2019). Rehabilitation needs of youth with arthrogryposis multiplex congenita: Perspectives from key stakeholders. *Disability and Rehabilitation*[Epub ahead of print], 1–7.

Eriksson, M., Bartonek, A., Pontén, E., & Gutierrez-Farewik, E. M. (2015). Gait dynamics in the wide spectrum of children with arthrogryposis: A descriptive study. *BMC Musculoskeletal Disorders*, *16*, 384. https://doi.org/10.1186/s12891-015-0834-5

Eriksson, M., Gutierrez-Farewik, E. M., Broström, 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, Å. (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

Eriksson, M., Villard, L., & Bartonek, A. (2014). Walking, orthoses and physical effort in a Swedish population with arthrogryposis. *Journal of Children's Orthopaedics*, *8*(4), 305–312. https://doi.org/10.1007/s11832-014-0597-9

Fassier, A., Wicart, P., Dubousset, J., & Seringe, R. (2009). Arthrogryposis multiplex congenita. Long-term follow-up from birth until skeletal maturity. *Journal of Children's Orthopaedics*, *3*(5), 383–390. https://doi.org/10.1007/s11832-009-0187-4

Fitzpatrick, R., Davey, C., Buxton, M.J., & Jones, D.R. (1998). Evaluating patient-based outcome measures for use in clinical trials. *Health Technology Assessment*, *2*(14), i-iv, 1–74

Friedlander, H. L., Westin, G. W., & Wood, W. L. (1968). Arthrogryposis multiplex congenita. *A review of forty-five cases*. *Journal of Bone and Joint Surgery. American Volume*, *50*, 89–112.

Gibson, D. A., & Urs, N. D. (1970). Arthrogryposis multiplex congenita. *Journal of Bone and Joint Surgery. British Volume (London)*, *52*(3), 483–493.

Green, A. D. L., Fixsen, J. A., & Lloyd-Roberts, G. C. (1984). Talectomy for arthrogryposis multiplex congenita. *Journal of Bone and Joint Surgery. British Volume (London)*, *66*(5), 697–699.

Gruel, C. R., Birch, J. G., Roach, J. W., & Herring, J. A. (1986). Teratologic dislocation of the hip. *Journal of Pediatric Orthopedics*, *6*(6), 693–702.

Guidera, K. J., & Drennan, J. C. (1985). Foot and ankle deformities in arthrogryposis multiplex congenita. *Clinical Orthopaedics and Related Research*, *194*, 93–98.

Hahn, G. (1985). Arthrogryposis. Pediatric review and habilitative aspects. *Clinical Orthopaedics and Related Research*, *194*, 04–14.

Hall, J. G. (1997). Arthrogryposis multiplex congenita: Etiology, genetics, classification, diagnostic approach, and general aspects. *Journal of Pediatric Orthopedics. Part B*, *6*(3), 159–166.

Hall, J. G., Aldinger, K. A., & Tanaka, K. I. (2014). Amyoplasia revisited. *American Journal of Medical Genetics Part A*, *164*(3), 700–730.

Hall, J. G., Reed, S. D., & Driscoll, E. P. (1983). Part I. Amyoplasia: A common, sporadic condition with congenital contractures. *American Journal of Medical Genetics*, *15*(4), 571–590. https://doi.org/10.1002/ajmg.1320150407

Hamdy, R., & Dahan-Oliel, N. (2016). Arthrogryposis. In S. Sabharwal (Ed.), *Pediatric lower limb deformities: Principles and techniques of management* (pp. 297–311), Switzerland: Springer International Publishing. https://doi.org/10.1007/978-3-319-17097-8_18

Hansen, O. M. (1961). Surgical anatomy and treatment of patients with arthrogryposis. *Journal of Bone and Joint Surgery. British Volume (London)*, *43B*, 855.

Himuro, N., Abe, H., Nishibu, H., Seino, T., & Mori, M. (2017). Easy-to-use clinical measures of walking ability in children and adolescents with cerebral palsy: A systematic review. *Disability and Rehabilitation*, *39*(10), 957–968. https://doi.org/10.1080/09638288.2016.1175036

Himuro, N., Abe, H., Nishibu, H., Seino, T., & Mori, M. (2017). Easy-to-use clinical measures of walking ability in children and adolescents with cerebral palsy: A systematic review. *Disability and Rehabilitation*, *39*(10), 957–968. https://doi.org/10.1080/09638288.2016.1175036

Ho, C. A., & Karol, L. A. (2008). The utility of knee releases in arthrogryposis. *Journal of Pediatric Orthopedics*, *28*(3), 307–313. https://doi.org/10.1097/BPO.0b013e3181653bde

Hoffer, M. M., Swank, S., Eastman, F., Clark, D., & Teitge, R. (1983). Ambulation in severe arthrogryposis. *Journal of Pediatric Orthopedics*, *3*(3), 293–296.

Holsbeeke, L., Ketelaar, M., Schoemaker, M. M., & Gorter, J. W. (2009). Capacity, capability, and performance: Different constructs or three of a kind? *Archives of Physical Medicine and Rehabilitation*, *90*(5), 849–855. https://doi.org/10.1016/j.apmr.2008.11.015

Hsu, L. C. S., Jaffray, D., & Leong, J. C. Y. (1984). Talectomy for club foot in arthrogryposis. *Journal of Bone and Joint Surgery. British Volume (London)*, *66*(5), 694–696.

Huurman, W. W., & Jacobsen, S. T. (1985). The hip in arthrogryposis multiplex congenita. *Clinical Orthopaedics and Related Research*, *194*, 81–86.

Janicki, J. A., Narayanan, U. G., Harvey, B., Roy, A., Ramseier, L. E., & Wright, J. G. (2009). Treatment of neuromuscular and syndrome-associated (nonidiopathic) clubfeet using the Ponseti method. *Journal of Pediatric Orthopedics*, *29*(4), 393–397. https://doi.org/10.1097/BPO.0b013e3181a6bf77

Jones, T., Miller, R., Street, J. T., & Sawatzky, B. (2019). Validation of the Oswestry disability index for pain and disability in arthrogryposis multiplex congenita. *Annals of Physical and Rehabilitation Medicine*, *62*(2), 92–97.

Kimber, E. (2009). Arthrogryposis. In *Causes, consequences and clinical course in amyoplasia and distal arthrogryposis*. Institute of Clinical Sciences, Department of Pediatrics, Sweden: University of Gothenburg, Gothenburg.

Kowalczyk, B., & Felus, J. (2015). Treatment of foot deformities in arthrogryposis multiplex congenita. *JBJS Reviews*, *3*(6), pii. https://doi.org/10.2106/JBJS.RVW.N.00103

Kowalczyk, B., & Feluś, J. (2016). Arthogryposis: 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

Kowalczyk, B., & Lejman, T. (2008). Short-term experience with Ponseti casting and the Achilles tenotomy method for clubfeet treatment in arthrogryposis multiplex congenita. *Journal of Children's Orthopaedics*, *2*(5), 365–371. https://doi.org/10.1007/s11832-008-0122-0

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

Lampasi, M., Antonioli, D., & Donzelli, O. (2012). Management of knee deformities in children with arthrogryposis. *Musculoskeletal Surgery*, *96*(3), 161–169. https://doi.org/10.1007/s12306-012-0218-z

LeBel, M. E., & Gallien, R. (2005). The surgical treatment of teratologic dislocation of the hip. *Journal of Pediatric Orthopaedics. Part B*, *14*(5), 331–336.

Lloyd-Roberts, G. C., & Lettin, A. W. (1970). Arthrogryposis multiplex congenita. *Journal of Bone and Joint Surgery. British Volume (London)*, *52*, 494–508.

Mead, N. G., Lithgow, W. C., & Sweeney, H. J. (1958). Arthrogryposis multiplex congenita. *The Journal of Bone and Joint Surgery. American Volume*, *40-A*(6), 1285–1309.

Menelaus, M. B. (1971). Talectomy for equinovarus deformity in arthrogryposis and spina bifida. *Journal of Bone and Joint Surgery. British Volume (London)*, *53*, 648–673.

Mensch, S. M., Rameckers, E. A., Echteld, M. A., & Evenhuis, H. M. (2015). Instruments for the evaluation of motor abilities for children with severe multiple disabilities: A systematic review of the literature. *Research in Developmental Disabilities*, *47*, 185–198. https://doi.org/10.1016/j.ridd.2015.09.002

Morcuende, J. A., Dobbs, M. B., & Frick, S. L. (2008). Results of the Ponseti method in patients with clubfoot associated with arthrogryposis. *The Iowa Orthopaedic Journal*, *28*, 22–26.

Moroney, P. J., Noel, J., Fogarty, E. E., & Kelly, P. M. (2012). A single-center prospective evaluation of the Ponseti method in nonidiopathic congenital talipes equinovarus. *Journal of Pediatric Orthopedics*, *32*(6), 636–640. https://doi.org/10.1097/BPO.0b013e31825fa7df

Mosca, V. M. (2001). Limited posteromedial clubfoot release. In *Staheli LT: Practice of pediatric orthopedics* (p. 388). Philadelphia, PA: Lippincott Williams & Wilkins.

Niki, H., Staheli, L. T., & Mosca, V. S. (1997). Management of clubfoot deformity in amyoplasia. *Journal of Pediatric Orthopedics*, *17*(6), 803–807.

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

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.

Pirani, S., Outerbridge, H. K., Sawatzky, B., & Stothers, K. (1999). A reliable method of clinically evaluating a virgin clubfoot evaluation. Paper presented at the Societé Interationale de Chirurgie Orthopédique et de Traumatologie Congress, Sydney, Australia.

Ponten, E. (2015). Management of knee in AMC. *Journal of Children's Orthopaedics*, *9*(6), 465–472. https://doi.org/10.1007/s11832-015-0695-3

Robertson, G. G., Williamson, A. P., & Blattner, R. (1955). A study of abnormalities in early chick embryos inoculated with Newcastle disease virus. *The Journal of Experimental Zoology*, *129*(1), 5–43.

Rocha, L. E., Nishimori, F. K., Figueiredo, D. C., Grimm, D. H., & Cunha, L. A. (2010). Open reduction of hip dislocation in patients with arthrogryposis multiplex congenita—An anteromedial approach. *Revista Brasileira de Ortopedia*, *45*(5), 403–408. https://doi.org/10.1177/230949900401200102

Sarwark, J. F., MacEwen, G. D., & Scott, C. I., Jr. (1990). Amyoplasia (a common form of arthrogryposis). *The Journal of Bone and Joint Surgery. American Volume*, *72*(3), 465–469.

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.

Sodergard, J., & Ryoppy, S. (1994). Foot deformities in arthrogryposis multiplex congenita. *Journal of Pediatric Orthopedics*, *14*(6), 768–772.

Song, K. (2017). Lower extremity deformity management in amyoplasia: When and how. *Journal of Pediatric Orthopedics*, *37*, s42–s47. https://doi.org/10.1097/BPO.0000000000001030

St Clair, H. S., & Zimbler, S. (1985). A plan of management and treatment results in the arthrogrypotic hip. *Clinical Orthopaedics and Related Research*, *194*, 74–80.

Staheli, L. T., Chew, D. E., & Elliott, J. S. (1987). Mosca vs. management of hip dislocations in children with arthrogryposis. *Journal of Pediatric Orthopedics*, *7*(6), 681–685.

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

Szoke, G., Staheli, L. T., Jaffe, K., & Hall, J. G. (1996). Medial-approach open reduction of hip dislocation in amyoplasia-type arthrogryposis. *Journal of Pediatric Orthopedics*, *16*(1), 127–130.

van Bosse, H. J. (2015). Syndromatic feet: Arthrogryposis and myelomeningocele. *Foot and Ankle Clinics of North America*, *20*, 619–644. https://doi.org/10.1016/j.fcl.2015.07.010

van Bosse, H. J. P., Marangoz, S., Lehman, W. B., & Sala, D. A. (2009). Correction of arthrogrypotic clubfoot with a modified Ponseti technique. *Clinical Orthopaedics and Related Research*, *467*(5), 1283–1293. https://doi.org/10.1007/s11999-008-0685-6

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*, s16–s23. https://doi.org/10.1097/BPO.0000000000001005

van Bosse, H. J., & Saldana, R. E. (2017). Reorientational proximal femoral osteotomies for arthrogrypotic hip contractures. *The Journal of Bone and Joint Surgery. American Volume*, *99*(1), 55–64.

Vanpaemel, L., Schoenmakers, M., van Nesselrooij, B., Pruijs, H., & Helders, P. (1997). Multiple congenital contractures. *Journal of Pediatric Orthopaedics*, *6*(3), 172–178.

Wada, A., Yamaguchi, T., Nakamura, T., Yanagida, H., Takamura, K., Oketani, Y., … Fujii, T. (2012). Surgical treatment of hip dislocation in amyoplasia-type arthrogryposis. *Journal of Pediatric Orthopaedics. Part B*, *21*(5), 381–385. https://doi.org/10.1097/BPB.0b013e328355d040

Wagner, L. V., Cherry, S. J., Sawatzky, B. J., Fafara, A., Elfassy, C., Eriksson, M., … Donohoe, M. (2019). Rehabilitation across the lifespan for children with arthrogryposis. *American Journal of Medical Genetics* [Epub ahead of print].

Wenger, D. R., Mubarak, S. J., Henderson, P. C., & Miyanji, F. (2008). Ligamentum teres maintenance and transfer as a stabilizer in open reduction for pediatric hip dislocation: Surgical technique and early clinical results. *Journal of Children's Orthopaedics*, *2*(3), 177–185. https://doi.org/10.1007/s11832-008-0103-3

Widmann, R. F., Do, T. T., & Burke, S. W. (2005). Radical soft tissue release of the arthrogrypotic clubfoot. *Journal of Pediatric Orthopaedics. Part B*, *14*(2), 111–115.

Williams, P. (1978). The management of arthrogryposis. *The Orthopedic Clinics of North America*, *9*(1), 67–88.

World Health Organization. (2001). International Classification of Functioning, Disability and Health (ICF). Retrieved from http://www.who.int/classifications/icf/en/.

World Health Organization. (2007). The International Classification of Functioning, Disability, and Health for Children and Youth (ICF-CY). Retrieved from http://www.who.int/classifications/icf/en/.

Yang, S. S., Dahan-Oliel, N., Montpetit, K., & Hamdy, R. C. (2010). Ambulation gains after knee surgery in children with arthrogryposis. *Journal of Pediatric Orthopaedics*, *30*(8), 863–869.

Yau, P. W., Chow, W., Li, Y. H., & Leong, J. C. (2002). Twenty-year follow-up of hip problems in arthrogryposis multiplex congenita. *Journal of Pediatric Orthopedics*, *22*(3), 359–363.

Zanudin, A., Mercer, T. H., Jagadamma, K. C., & van der Linden, M. L. (2017). Psychometric properties of measures of gait quality and walking performance in young people with cerebral palsy: A systematic review. *Gait & Posture*, *58*, 30–40. https://doi.org/10.1016/j.gaitpost.2017.07.005

Zimbler, S., & Craig, C. L. (1983). The arthrogrypotic foot plan of management and results of treatment. *Foot & Ankle*, *3*(4), 211–219.

How to cite this article: Hamdy RC, van Bosse H, Altiok H, et al. Treatment and outcomes of arthrogryposis in the lower extremity. *Am J Med Genet Part C*. 2019;181C:372–384. https://doi.org/10.1002/ajmg.c.31734