Orthopaedic care of the child with arthrogryposis: a 2020 overview

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Purpose of review
The orthopaedic treatment of children with arthrogryposis multiplex congenita has evolved steadily over the past two decades. Interrelated factors have spurred this on, including better appreciation of the functional potential of persons with arthrogryposis, development of newer procedures specific for the arthrogrypotic deformities, and outcomes studies that provide understanding of the overall capabilities of adults with arthrogryposis and follow-up to determine which treatments were beneficial and which were not. This article briefly sketches out of some of these advances and indicates areas that need further development.

Recent findings
Outcome studies show that the majority of adults with arthrogryposis are ambulatory but less than half are fully independent. Adults frequently experience ongoing pain, particularly foot and back pain, limiting ambulation and standing. Advancements in the upper extremity treatment include improving elbow function, wrist repositioning, and improving thumb positioning. In the lower extremities, correction of hip and knee contractures leads to improved ambulatory potential, and treating clubfeet with serial casting decreases poor outcomes.

Summary
Clinical evaluation, both physical examination and assessment of the patient's needs, are important in directing treatment in arthrogryposis. Further outcomes studies are needed to continue to refine procedures and define the appropriate candidates.

Keywords
arthrogryposis, arthrogryposis multiplex congenita, clubfoot, congenital contracture syndrome, elbow contractures, hip contractures, knee contractures, wrist contractures

INTRODUCTION

The term arthrogryposis multiplex congenita, or simply arthrogryposis, describes a collection of conditions with congenital contractures of two or more joints in multiple body areas [1]. The estimated incidence is between 1 : 3000 to 1 : 5000 live births, with some 400 underlying diagnoses identified so far [2,3]. The underlying commonality is a lack of intrauterine movement (fetal akinesia), resulting in joint fibrosis and stiffness [4]. Heterogeneity of the causative conditions has led to ongoing international efforts to craft a precise definition of arthrogryposis and meaningful classification systems [5▪,6,7▪]. The past 10–12 years have seen a huge growth in understanding and interest in treating both children and adults with arthrogryposis. Diagnoses may be difficult and costly to determine [8▪,9▪]; therefore, some studies group all people with arthrogryposis together, whereas others may focus solely on a particular diagnosis, such as amyoplasia, the most common cause of arthrogryposis.

OUTCOMES STUDIES

A recognized need in discussing treatment protocols for children with arthrogryposis is understanding their late effects [10▪,11▪▪,12▪▪]. Recent studies have detailed outcomes in adults with arthrogryposis. Ambulation abilities are relatively high (49–88%), although many use wheelchairs in the community or for longer distances; some adults report losing the ambulation ability they had as children [13▪–18▪]. Many drive automobiles (48–86%), with only occasional control modifications [13▪,16▪]. Only 32–50% are fully independent in their activities of daily living, usually related to upper extremity function

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KEY POINTS

- The majority of adults with arthrogryposis are ambulatory, although less than half are fully independent in their daily lives.
- Pain is a common concern of adults with arthrogryposis, particularly related to their spine and ankles/feet, which for some impacts standing and walking activities.
- Prenatal workup can help prepare parents for their baby's future needs, but the rate of prenatal detection is unfortunately still low.
- Advancements in upper extremity care include better patient selection and refinement of surgical technique, particularly in gaining elbow motion.
- Advancements in lower extremity care include regular use of Ponseti casting for clubfeet, and development of new techniques or advancement of previous procedures in treating hip and knee contractures, or congenital hip dislocations.
- Arthrogrypotic spine deformities are still largely unstudied, although there is early understanding of the utility of spine casting and nonfusion spinal instrumentation techniques.

[14▪,16▪]. The vast majority (95–98%) had undergone orthopaedic procedures, on average 7–10 procedures, but as many as 45 or 70, most occurring during childhood [13▪–19▪].

Some 75–91% of adults describe pain as a common problem [13▪–15▪,17▪,20▪]. Pain affects feet/ankles, knees, spine, and less often the jaw and the upper extremities, is sometimes chronic, even from childhood, and is often activity limiting. Jones *et al.* [21▪] validated the Oswestry Disability Index for arthrogryposis, finding that pain of the low back and lower extremities was the most disabling, particularly for standing and walking activities.

These studies should inform us that most children with arthrogryposis have a significant potential for ambulation and independent living as adults. Treatment programs should be relatively aggressive, to help children realize their full potential, yet judicious and individualized, to mitigate the risk of overtreatment. These treatments should accentuate goals of decreasing pain in adults, including fewer surgical interventions for foot deformities, and emphasis on standing posture to preclude back pain.

PRENATAL CONSULTATION

Occasionally pediatric orthopaedists are asked for prenatal evaluations related to ultrasound or MRI findings. These can be difficult discussions because of the uncertainties involved, especially if the parents are struggling with considerations to terminate the pregnancy. Giving advice on pregnancy termination is outside the purview of most pediatric orthopaedists, but understanding the broader picture of arthrogryposis is helpful. Fetal and neonatal mortality is common. One study found that of 183 arthrogrypotic pregnancies, 3 were spontaneous losses before 20 weeks' gestation, 12 pregnancies were terminated, 18 were stillborn, and 50 died (most in the first month of life), for a total mortality rate of 45% [22]. Early prognostic ultrasound findings include fetal hydrops (fluid accumulation in two or more compartments, such as ascites, pericardial or pleural effusions, and skin edema), cystic hygroma (fluid-filled sac secondary to lymphatic blockage) or nuchal edema/translucency (cystic hygroma of the posterior neck region); later signs include pulmonary hypoplasia, polyhydramnios and absent stomach filling, all evidence of a lack of intrauterine breathing and swallowing [23,24▪▪,25▪,26▪▪]. Fetal MRI may demonstrate brain malformations or insufficient lung volumes [24▪▪,25▪]. This information provides the orthopaedist with an indication of involvement severity.

In addition to providing information on postnatal orthopaedic treatment, orthopaedists can also recommend evaluation for fetal cervical hyperextension (FCH) on ultrasound, especially if the fetus is breech [27▪]. If FCH is present, a careful Caesarian delivery is indicated, to avoid trauma to the cervical spinal cord.

Unfortunately, the rate of prenatal detection of arthrogryposis is still low, as current ultrasound protocols only diagnose approximately 25% of fetuses with arthrogryposis before 24 weeks gestation [28,29▪]. Yet, studies show that competent ultrasonographers can detect nearly half of prenatally detectable structural abnormalities by 12–13 weeks gestation if following proper protocols [30▪]. Awareness to these issues should lead to the institution of better prenatal screening protocols to identify at risk pregnancies earlier [29▪].

UPPER EXTREMITY

Outcomes studies have driven changes in treatment of the upper extremities of children with arthrogryposis over the past several years, both in philosophy and practice. For example, patients describe the practice of surgically creating a flexed and contralateral straight elbow, in order to have 'one eating and one toileting arm' as inappropriate [16▪].

Shoulder

Improvement in shoulder function in children with arthrogryposis continues to be difficult, as there are no reliable procedures to increase shoulder strength or motion range [31▪▪,32▪]. Instead, exoskeleton type devices have been developed, which support the weight of the arm against gravity [33▪,34▪,35]. These allow children to explore their environment and participate in more complex free play. The bulk of the devices can effect tolerance to use.

Internal rotational deformity of the upper arm, either from shoulder contracture or humeral torsion, positions the elbow axis into the sagittal plane, prohibiting activities in front of the body. The humeral derotational osteotomy is effective in redirecting the palm towards the midline [36,37]. The osteotomy is most commonly positioned distally, which allows for using the same incision as for a posterior elbow release. Average derotation of 90° is recommended, and if a posterior elbow release is to be done concurrently, rigid fixation of the osteotomy is imperative for early elbow range of motion exercise [31▪▪,36,37]. The high rate of periimplant fractures make hardware removal necessary, and repeat surgery is occasionally required for the poorly understood phenomena of rotation recurrence [31▪▪,32▪,37].

Elbow

Currently, there are no described procedures to address elbow flexion contractures. The emphasis instead has been on improving elbow flexion, both passive and powered, in children with stiff elbow extension contractures. The goal is to improve self-feeding and grooming by providing at least 90° of elbow flexion. A simple release is performed by lengthening the triceps tendon (by W or VY-plasty), elbow capsulotomy, and anterior transposition of the ulnar nerve [32▪,36]. Performing the release before 2 years of age possibly improves the overall passive total arc of elbow motion at followup, compared with children 2–3 years old, and older [38▪].

Adults with good passive elbow motion are more likely to be independent compared with those without, but active elbow flexion is associated with increased ability to eat/drink and dress the upper body [16▪,19▪]. Procedures to power elbow flexion are applicable to only a select group of patients. The elbow should have 90° of passive flexion, and have suitable recruitable muscles for transfer; an MRI can help determine if a muscle has appropriate bulk [31▪▪,32▪]. Options for transfer include the latissimus dorsi, the pectoralis major, a free gracilis transfer or the long head of the triceps – if the lateral head is also robust. If a posterior elbow release is performed at the same time as the long head of triceps is transferred, there is a risk of losing elbow extension [32▪]. A bipolar latissimus dorsi transfer has been described, with the proximal end attached to the coracoid process, and the distal end is weaved through the biceps tendon and attached to the bicipital tuberosity [39▪]. The active elbow range was 98° at follow-up, with the ability to flex the elbow against resistance in nearly all patients. A technique is described as transferring the median nerve to the musculocutaneous nerve in infants shortly after birth (around 3–4 months) in three patients [40▪]. A healthy biceps muscle need be demonstrated by ultrasound, and as well as active finger/wrist flexion, to ensure that recipient and donor are appropriate. At an average of 4 years' follow-up, average biceps strength was M3.

Wrist

The common wrist deformity is flexion and ulnar deviation. When the wrist can be actively extended to neutral, further extension can be attained by a release of the volar fascia with lengthening or tenotomy of the wrist flexors, paired with a transfer of the extensor carpi ulnaris (ECU) to the extensor carpi radialis brevis [31▪▪]. If the wrist cannot reach neutral extension actively or passively, a carpal wedge osteotomy repositions the hand well [31▪▪,32▪,41▪]. The biplanar osteotomy both extends the wrist and corrects the ulnar deviation, usually with a volar release and ECU transfer. There is approximately 5° to 10° loss of total arc of motion, but this is more than compensated by the improved hand positioning [41▪].

Thumb

In the clasped thumb, or thumb-in-palm deformity, the first metacarpophalangeal (MCP) joint lacks extension and is unstable, the first web space is narrowed with an associated skin deficiency, and the underlying muscles and fascia are contracted. Treatment includes a modified dorsal rotation advancement flap to augment skin of the first web space (Abdel-Ghani flap), releases of the underlying tight structures, possible fractional lengthening of the flexor pollicis longus, and chondrodesis of the thumb MCP joint [31▪▪,32▪,42▪]. This positions the thumb out of the plane of the palm, providing a post against which to pinch, and improving the hand cosmesis [41▪]. The relapse risk is high if the extensor pollicis longus is not functional and no transferable tendon is available for thumb extension [32▪]. The chondrodesis nonunion rate is approximately 10%, often leading to instability and needing revision surgery [41▪].

LOWER EXTREMITY

Over the past two decades, treatment of children with lower extremity contractures has become more interventionistic. No longer is it acceptable to relegate a child with moderate-to-severe contractures to lifetime wheelchair use without careful assessment of their likelihood of ambulation. Most patients have the potential to ambulate, and even those who have resorted to wheelchair use as adults appreciate standing for transfers or ambulating short distances within the home.

Hip

Congenital hip dislocations in arthrogryposis are teratologic, occurring earlier in pregnancy, and therefore, are stiffer with higher displacement. Nonsurgical methods of relocation are not effective. Historically, the concerns have been that surgical relocation of such hips would lead to stiffness, but follow-up studies show that only an average of 4° of flexion–extension arc was lost following open reduction in comparison to preoperative motion [43▪▪]. Both the anterolateral and medial approaches have been described for arthrogrypotic hip dislocations [43▪▪,44▪], and stability can be enhanced by using the ligamentum teres to tether the femoral head within the acetabulum, or provisionally pinning the femoral–acetabular joint (Fig. 1) [45,46]. Avascular necrosis occurred in 24% of hips, 80% of which were older than 3 years of age, but did not seem to affect hip motion [43▪▪].

Contractures of the hip can vary from mild and moderate uniplanar to severe multiplanar. Often, the contracted conjoin tendon of the sartorius and tensor fascia lata can be palpated just inferior to the anterior superior iliac spine (ASIS), and in cases of isolated hip flexion contractures, a percutaneous release of that structure may significantly improve hip extension [43▪▪,44▪,47]. The more severe contractures are multiplanar, with elements of flexion, abduction, and external rotation. The reorientational osteotomy of the proximal femur repositions the lower limb in line with the body, leaving the femoral head in its original position within the acetabulum [43▪▪,44▪,47]. In this way, the existing motion of the hip is preserved, the total arc of motion is unchanged, but the limb itself is oriented more functionally (Fig. 2). In one study, ambulation increased from 9% pretreatment to 55% at follow-up, with another 31% still walker dependent [47].

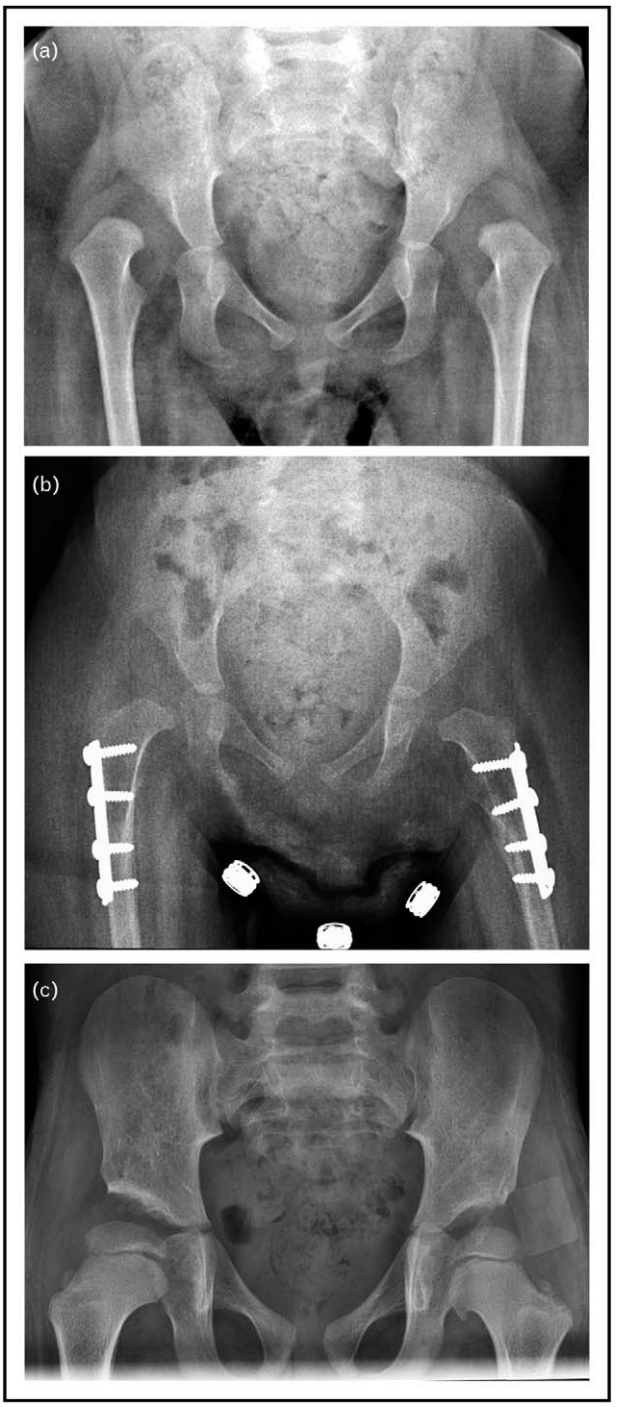

FIGURE 1. Twenty-month-old boy with arthrogryposis, with bilateral hip dislocations: (a) Anteroposterior (AP) pelvis radiograph preoperatively, demonstrating bilateral hip dislocations; (b) AP pelvis radiograph following bilateral hip open reduction with ligamentum teres tethering of the femoral head, and bilateral femoral shortening osteotomies; (c) follow-up AP pelvis radiograph at 6 years of age. The patient ambulates independently.

Knee

Knee flexion contractures can be classified as mild (<30°), moderate (30–50°) and severe (>60°). For the mild and moderate ones, growth guidance with femoral anterior distal hemiepiphysiodesis (FADHE) provides for gradual correction with growth [48],

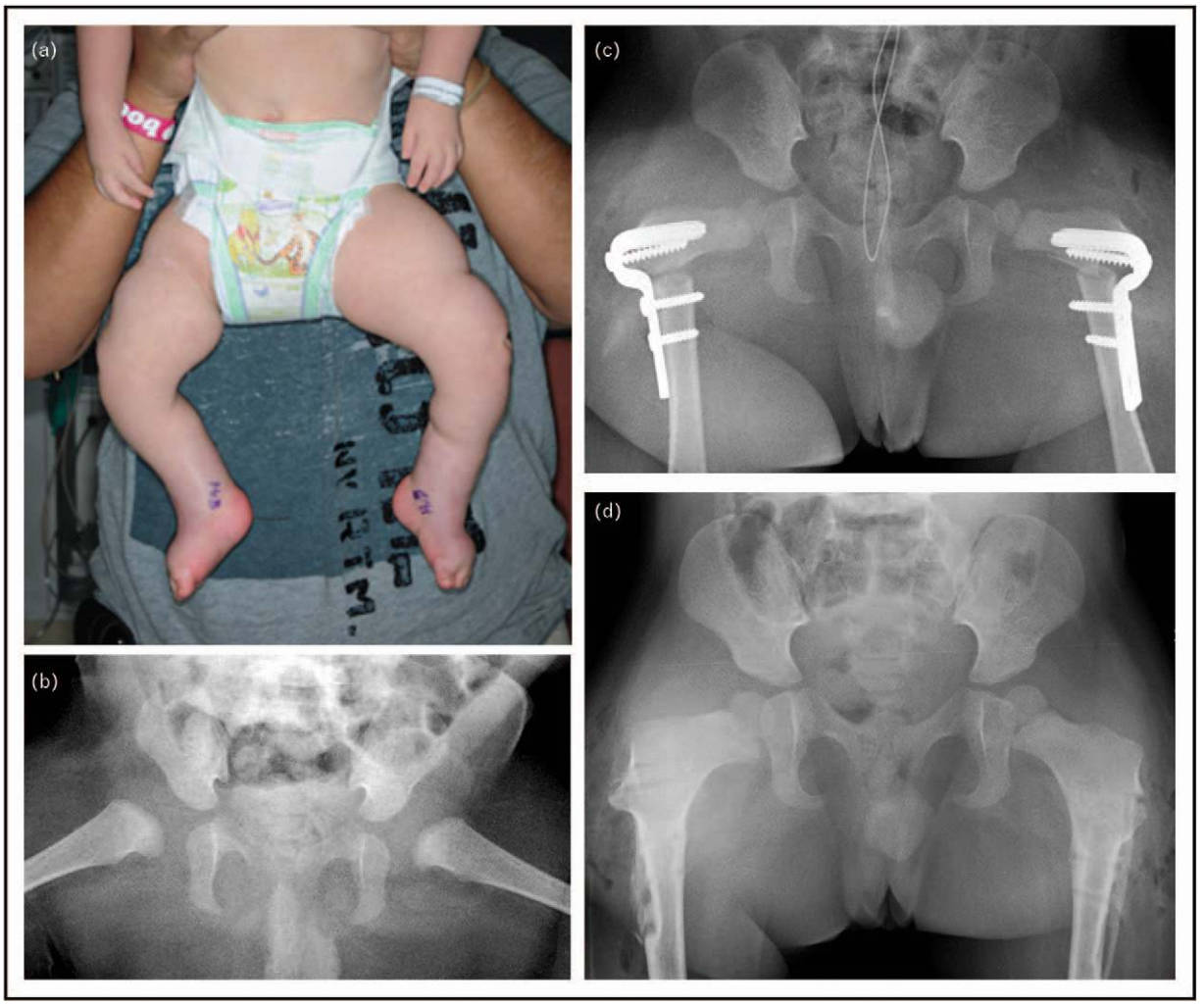

FIGURE 2. Sixteen-month-old boy with arthrogryposis and bilateral hip contractures: (a) clinical photograph of patient's hip contractures. He had 70° hip flexion, 45° adduction, and 20° external rotation contractures. (b) Anteroposterior (AP) pelvis radiograph preoperatively, demonstrating contractures; (c) AP pelvis radiograph immediately postoperatively, following bilateral proximal femoral reorientational osteotomies. Note that the proximal femur is in essentially the same position as in the preoperative film; (d) AP pelvis radiograph after removal of hip hardware removal, 15 months postoperatively. Note the significant proximal femoral remodeling that has already occurred.

supplemented with a posterior release of the knee for the moderate contractures (Fig. 3). The severe contractures are unlikely to fully respond to FADHE, instead a knee spanning external fixator with posterior knee release allows for gradual extension of the joint [49]. Proper placement of the fixator hinge, in line with the knee rotational axis, is important to prevent subluxation, and an initial distraction of the joint avoids joint impingement at extension. Contracture recurrence with both FADHE and joint distraction is common; patients are encouraged to wear a knee–ankle–foot orthosis (KAFO) at nighttime for many years to limit that process. Distal femoral extension osteotomies allow for acute and full correction, but is best indicated for patients nearing skeletal maturity because of a high relapse rate from physeal remodeling [50].

Extension contractures are less common, and initially are less problematic, since they do not hinder ambulation. Eventually they can make sitting difficult, for example, in automobiles. In infants and young children, if serial casting fails, then a percutaneous or miniopen rectus femoris tenotomy is indicated [51]. In more severe contractures and in older children, a Judet quadricepsplasty, possibly with a femoral shortening osteotomy, will have better success [52]. The child will likely need to ambulate with more extensive orthoses (KAFO or floor reaction AFO) for an extended period of time, as they regain strength and balance.

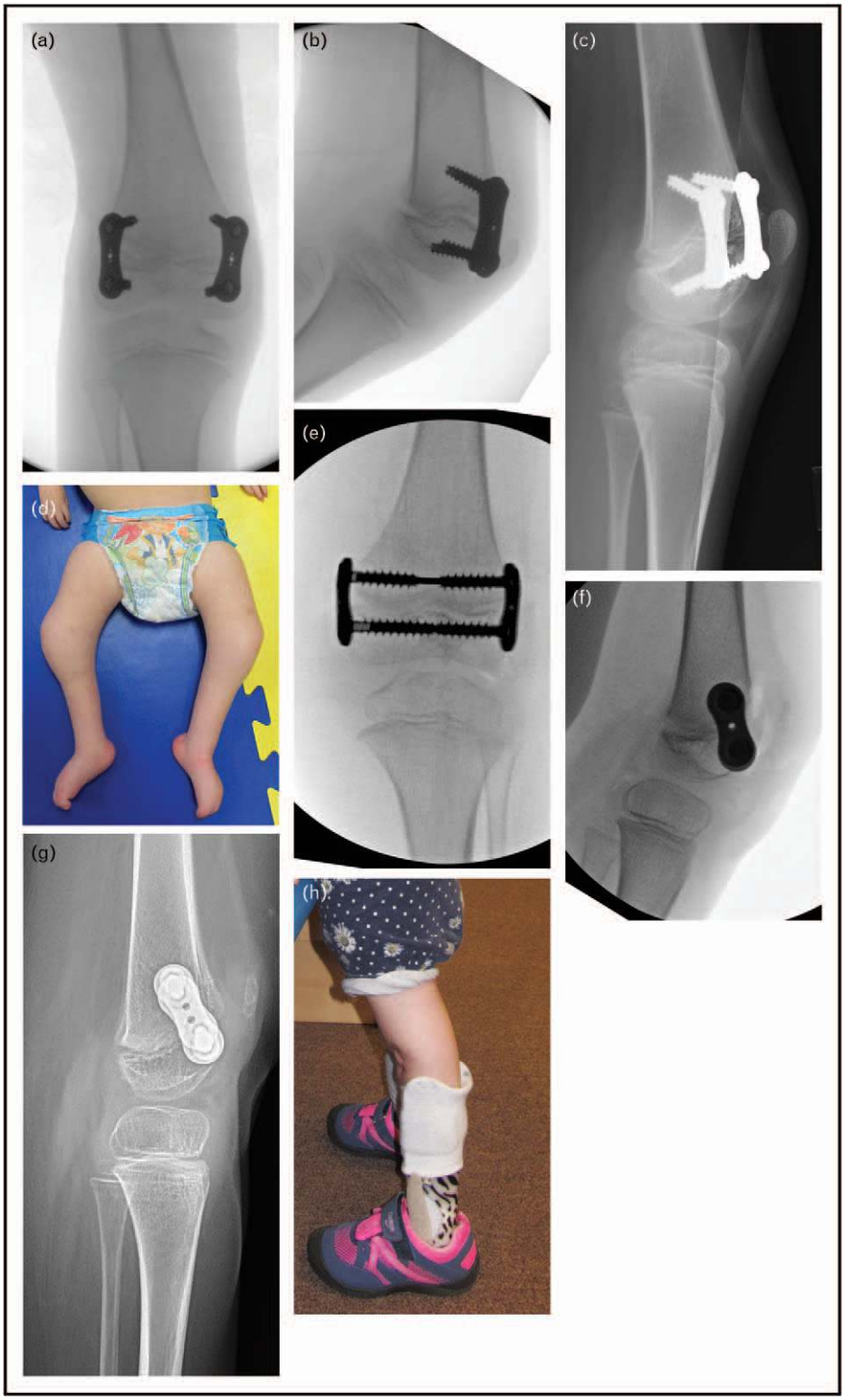

FIGURE 3. Anterior distal femoral hemiepiphysiodesis: (a) immediate postoperative AP radiograph of the right knee of an 8-year-old boy with arthrogryposis, after placement of medial and lateral physeal tethering plates for a 20° knee flexion contracture; (b) immediate postoperative lateral radiograph of right knee; (c) lateral radiograph of right knee 12 months later, demonstrating divergence of the fixation screws, and correction of the flexion contracture. (d) Clinical photograph of a 3-year-old girl with arthrogryposis, demonstrating her 45° and 25° left knee flexion contractures; (e) immediate postoperative AP left knee radiograph following bilateral anterior distal femoral hemiepiphysiodesis with the physeal bridge technique at 6 years old; (f) immediate postoperative lateral left knee radiograph; (g) follow-up lateral radiograph of left knee 5 months postoperative, demonstrating significant physeal obliquity; (h) clinical photograph demonstrating a fully extended knee.

Foot

The typical arthrogrypotic foot deformities are the clubfoot or the congenital vertical talus. Arthrogrypotic clubfeet have a penchant for relapse, regardless of the treatment used. Relapses after surgical correction are often stiffer, more painful, and more difficult to correct. Increasingly arthrogrypotic clubfeet are treated with the Ponseti method [53,54,55▪], where relapses can be treated with repeat casting. In cases of severe equinus, an Achilles tenotomy is done early, either initially or in mid treatment, to unlock the calcaneus from the posterior tibia (Fig. 4). Bracing is an important part of the treatment. The standard foot-abduction brace (FAB) is effective for the idiopathic clubfoot, but less so for the arthrogrypotic one, probably as the arthrogrypotic patient lacks active ankle motion, and unlike a typically developing child, does not spontaneously kick. We have noticed a high prevalence of external tibial torsion associated with arthrogrypotic clubfeet, making it nearly impossible to abduct the brace suitably to position the foot relative to the ankle. Instead, we use a leaf spring ankle–foot orthosis (AFO), pushing the ankle into valgus with a lateral supramalleolar pad and an ankle strap that originates inside laterally, and correcting the forefoot adductus with a medial forefoot pad. Removable nighttime straps dorsiflex the ankle to stretch the hindfoot.

The equinocavus foot is a variant of the classic clubfoot with has less heel varus and forefoot adductus, but pronounced midfoot cavus and a transverse plantar crease [44▪,55▪]. It closely resembles the atypical clubfoot described by Ponseti *et al.* [56] and similarly does poorly with standard Ponseti casting, developing into a irrecoverable valgus-abductus foot. The deformity does respond well to the same casting method that Ponseti described for

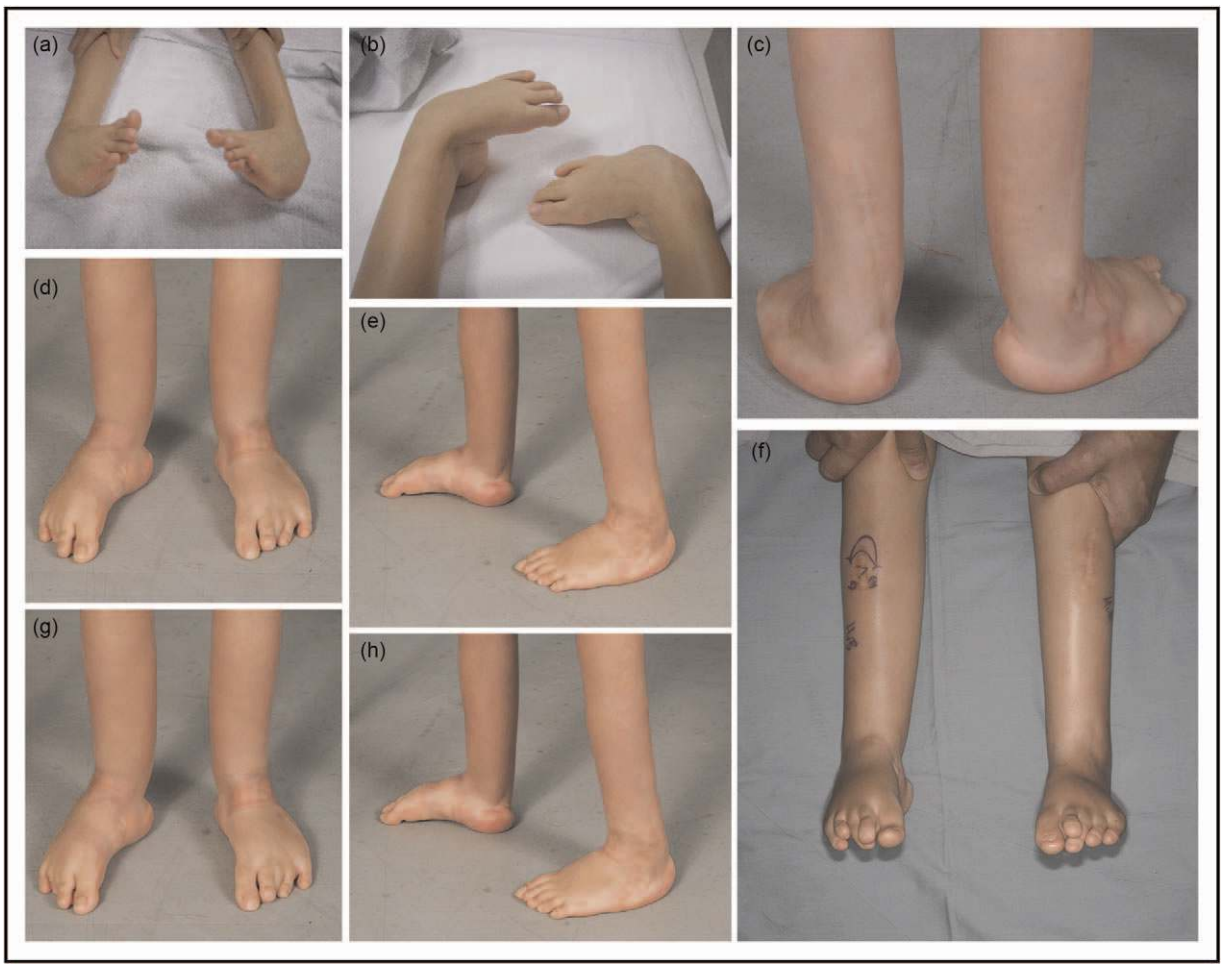

FIGURE 4. Nine-year-old girl with bilateral arthrogrypotic clubfeet, with relapses after prior surgery at unknown age: (a and b) pretreatment clinical photographs of both feet; (c–e) clinical views of both feet 14 months later. Treatment was eight sets of Ponseti style serial casts, midtreatment percutaneous Achilles tenotomies, and another eight sets of casts, over the course of 4 months; (f–h) follow-up at 16 years of age. The patient only complains of a prominent base of the fifth metatarsal that was treated by resecting the prominence.

the atypical clubfoot, where the forefoot is dorsiflexed relative to the midfoot, with minimal to no abduction [55▪,56].

Dobbs *et al.* revolutionized the treatment of the congenital vertical talus, with a 'reverse clubfoot' casting, which emphasized plantarflexion, forefoot supination and adduction [57,58]. A pinning of the talonavicular joint and possibly an Achilles tenotomy is required prior to the last cast, although I have always needed to perform an open reduction of the joint through a small incision prior to pinning. An AFO maintains the foot position, molded to correct hindfoot valgus and forefoot adductus, the ankle strap originating inside medially.

SPINE

The prevalence of spinal deformity in arthrogryposis is about 23% [59▪]. The presence of fetal cervical hyperextension should be assessed prenatally, in case a careful Caesarian section delivery is indicated to prevent cervical spine injury [27▪]. Many of the curves are present at birth, reflecting the baby's intrauterine position. These are not 'congenital scoliosis' in the common use of the term, as there are no vertebral malformations; rather they can be described as prenatal curves. Mehta casting can decrease the size of curves, or slow progression, but corrects the scoliosis [59▪,60]. Nonfusion spinal instrumentation with expandable implants is a valuable treatment method for the growing spine, but spine stiffness and autofusion limits the effective treatment time to about 5 years on average, so delaying surgery can allow for more overall growth [59▪,61]. Pulmonary dysfunction is not only related to the scoliosis, but also to hypokyphosis, and increased BMI [62▪]. Spinal fusion for arthrogrypotic scoliosis is very effective; in general the fusion needs to be longer than in idiopathic curves (14 versus 12 levels), with a lower expected percentage correction rate (44 versus 71%) [63▪]. The reoperation risk is 7% at 1 year and 29% at 4 years, mostly for infection (early) and pseudoarthosis (late) [64▪].

CONCLUSION

The fundamentals of treatment of the child with arthrogryposis are changing dramatically, both because of an understanding of the potential most of these children have, and to the development of more effective procedures. An international community of practitioners devoted to treating adults and children with arthrogryposis is developing, and projects are underway to better describe outcomes [10▪,18▪,65▪]. This should allow the next generation of those born with arthrogryposis to be treated more specifically for their individual needs, and be able to participate more fully in society.

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Conflicts of interest

There are no conflicts of interest.

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Papers of particular interest, published within the annual period of review, have been highlighted as:

- ▪ of special interest
- ▪▪ of outstanding interest

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9. ▪ Wallach E, Walther-Louvier U, Espil-Taris C, *et al.* Arthrogryposis in children: Etiological assessments and preparation of a protocol for etiological investigations. Arch Pediatr 2018; 25:322–326.

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10. ▪ Elfassy C, Darsaklis VB, Snider L, *et al.* Rehabilitation needs of youth with arthrogryposis multiplex congenita: Perspectives from key stakeholders. Disabil Rehabil 2019; DOI: 10.1080/09638288.2018.1559364 [Epub ahead of print]

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