

Review Article

A review of the orthopedic interventions and functional outcomes among a cohort of 114 children with arthrogryposis multiplex congenita

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Abstract.

PURPOSE: Arthrogryposis multiplex congenita (AMC) refers to a large heterogeneous group of conditions involving joint contractures in two or more different areas of the body. Contractures can lead to decreased range of motion and strength, and affect ambulation and autonomy. The aim of this study was to describe the orthopedic interventions and functional outcomes of a large cohort of children with AMC followed in a pediatric orthopedic center.

METHODS: A retrospective chart review of all children diagnosed with AMC followed at Shriners Hospital for Children – Canada (SHC) between January 1979 and July 2016 was conducted. One hundred twenty patients were identified, of whom six were excluded due to misdiagnosis or insufficient chart information. One hundred fourteen were retained. Patient demographics, AMC classification, comorbidities, operative and non-operative treatments received as well as community ambulation status, level of autonomy in self-care and transfers at latest follow-up were recorded.

RESULTS: There were 54 males and 60 females with a mean age at last clinic visit of 10 years 3 months. Amyoplasia and distal arthrogryposis (DA) were equally represented in our sample, 47 (41.2%) and 49 (43.0%) participants respectively, with the category *Other* comprising the remaining 18 (15.8%) participants. Children with DA had less involvement of the proximal joints than those in the two other groups. Contractures and deformities of the foot and ankle were the most prevalent, affecting 91.5% with Amyoplasia, 85.7% with DA and 83.3% in the *Other* category. Contractures of the shoulder and elbow were more common among individuals with Amyoplasia and those categorized *Other* than those with DA. In terms of walking ability, 98% of participants with DA were independent ambulators. Walking ability varied among the *Other* participants. Similarly, most children with DA were independent in self-care and transfers at the most recent follow-up.

CONCLUSION: The relatively large sample size of this study allowed for a better insight into the challenges associated with AMC management. These findings demonstrated the need for genetic testing to provide accurate diagnosis and classification, along with the use of standardized outcome tools to measure effectiveness of interventions. As AMC is rare, multi-site prospective studies are needed to improve research opportunities, develop functional measures specific to AMC and disseminate findings on a wider scale.

Keywords: Arthrogryposis multiplex congenita, surgical management, rehabilitation, functional outcome, ambulation, self-care

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1. Introduction

Arthrogryposis multiplex congenita (AMC) refers to a large heterogeneous group of conditions involving joint contractures in two or more different areas of the body [1]. AMC is a congenital condition which generally results from reduced movement of the foetus during pregnancy [2]. Fetal akinesia can in turn be caused by an array of environmental or genetic factors, or likely a combination of the two [3]. Prevalence of AMC is 1:3000–5000 newborns (thus considered to be rare [4]) and roughly affects males and females equally [1,5].

Contractures in one or more areas of the body commonly restrict ambulation and autonomy in patients. As clinical presentation and functional prognosis can vary widely depending on the precise diagnosis, general agreement is that management of this orthopedic disorder should be individualized, multidisciplinary and start early in life [6–9].

Main goals in the management of AMC include achieving mobility and autonomy through the least invasive therapies to promote developmental milestones and participation in daily activities. Conservative and surgical approaches are used to reduce contractures at affected joints to enable ambulation and function in daily life [5]. In the lower extremities, goals include having adequate range of motion (ROM) to allow for walking, standing and sitting [10]. In the past, surgical management has often been the treatment of choice. Recently, less invasive surgical techniques and early rehabilitation services are being offered (e.g. Ponseti technique, early daily passive stretching, orthoses), reducing the average number of surgeries needed per child [6].

The literature on AMC has focused on genetic studies, classification systems, and single-site studies reporting surgical outcomes. The aim of this retrospective chart review was to describe the different phenotypes and associated diagnoses, clinical presentation, orthopedic interventions and functional status at latest follow-up of children with AMC who present to a pediatric orthopedic setting. A better understanding of the clinical and surgical needs of children with AMC will promote the development of treatment guidelines and align future research priorities.

2. Methods

A retrospective chart review of all children with AMC followed at the Shriners Hospital for Children – Canada (SHC) between January 1979 and July 2016

was conducted. Administrative and ethical approvals were obtained from the Shriners and the local institutional review board, respectively. Charts were retrieved and reviewed to identify patients who: (i) had a clinical manifestation of multiple congenital contractures, defined as the presence of two or more contractures in two different areas of the body, and (ii) had clinical and medical information from at least one outpatient visit. Charts of 120 participants were identified, of whom six were excluded due to misdiagnosis or insufficient chart information; therefore 114 children met the selection criteria and were included in the study.

Demographic information, date of last clinic follow-up, AMC classification, comorbidities as documented in the medical chart, joint involvement, and all orthopedic interventions (subcategorized as bony procedures, soft tissue releases or serial casting) were recorded. Ambulation, use of bracing and ambulation aid at any point of the rehabilitation treatment, and level of autonomy in activities of daily living (ADLs), specifically self-care and ability to transfer, at latest follow-up, were also recorded.

AMC classification was determined by the research team during the review of the medical chart using Hall and colleagues' recent classification based upon the child's cardinal features [11]. This classification system consists of three groups: *Amyoplasia*, *distal arthrogryposis* and *everything else*. For the purpose of this paper, we will refer to the *everything else* category as *Other*. We chose to use this latter classification system as it is the easiest to apply clinically when genetic testing is unavailable and is the most understandable for families and clinicians. *Amyoplasia* was defined as involvement in all four limbs, usually symmetric, with associated muscle abnormalities in the absence of an identified genetic diagnosis. *Distal arthrogryposis* was defined as two or more contractures of mainly the distal extremities (hands and feet), a confirmed genetic diagnosis, or an associated syndrome (e.g. Freeman-Sheldon syndrome, Escobar syndrome etc.). Cases which described the presence of pterygium, x-linked conditions, involvement of the central nervous system or other syndromic-type features in addition to multiple congenital contractures were categorized as *Other* [8].

Ambulation was defined as the ability to ambulate in the community and consisted of three categories as previously described [12]: community ambulation, ambulation with device, and non-ambulation. Community ambulation was defined as being able to ambulate for long distances in the community without any device. Ambulation with device was defined as ambulation with

Table 1
Joint involvement per AMC category

	Amyoplasia (n = 47)		Distal arthrogryposis (n = 49)		Other (n = 18)	
	Joint involvement	NS	Joint involvement	NS	Joint involvement	NS
Upper Extremities (UE)						
Shoulder	33 (70.2%)	0	7 (15.9%)	5	10 (58.8%)	1
Elbow	36 (76.6%)	0	13 (29.5%)	5	10 (58.8%)	1
Wrist and hand	37 (78.7%)	0	31 (70.4%)	5	11 (64.7%)	1
All UE joints affected	25 (55.5%)	2	6 (14.3%)	7	4 (23.5%)	1
UE involvement only	2 (4.3%)	0	3 (6.25%)	1	0 (0.0%)	1
Lower Extremities (LE)						
Hip	34 (74.5%)	0	25 (52.0%)	1	17 (94.4%)	0
Knee	39 (82.9%)	0	29 (60.4%)	1	16 (88.8%)	0
Ankle and foot	43 (91.5%)	0	42 (85.7%)	1	15 (83.3%)	0
All LE joints affected	30 (63.8%)	0	16 (33.3%)	1	10 (58.8%)	1
LE involvement only	6 (12.8%)	0	7 (16.6%)	7	3 (16.7%)	0

*NS = Not specified in chart.

a device such as a cane, crutches, or walker for long distances in the community, but they may ambulate without a device at home. Non-ambulation was defined as using a wheelchair for mobility.

A similar method was developed to define level of autonomy in transfers and self-care using three categories: independent (i.e., able to transfer/complete self-care with no assistance), independent with minimal assistance (i.e., able to transfer/complete self-care with minimal assistance from a person or device), and dependent (i.e., requires moderate to maximum assistance from a person or device).

As children typically develop independent walking by the age of two and autonomy in self-care by the age of seven, patients who were younger than the cut-offs were excluded from the description of ambulation and self-care/transfers abilities respectively.

For purposes of data analysis, an electronic case report form was created in a secure database. The de-identified data was extracted into Excel for statistical analysis using descriptive statistics (mean, median, and standard deviation).

3. Results

3.1. Demographics

There were 54 males (47.4%) and 60 females (52.6%). Mean age at last clinic follow-up was 10 years 3 months (range: 2 months to 21 years). All participants were referred to the pediatric orthopedic hospital for a consultation. Fifty-four patients (47%) were from Quebec, fifty-two patients (46%) were from other Canadian provinces and 8 patients (7%) were from outside Canada. The range of orthopedic services offered

included ongoing follow-up, second opinion, or targeted surgical intervention. Other services offered included rehabilitation, social services, genetic and nutrition counseling. Extent of services or follow-up varied according to availability of local expertise, family resources, and proximity.

3.2. AMC classification

Amyoplasia and *distal arthrogryposis* were equally represented in our sample, 47 (41.2%) and 49 (43.0%) participants respectively, with the category *Other* comprised of the remaining 18 (15.8%) participants. Presence of comorbidities, while not consistently documented in the medical chart, was reported in varying detail for all three AMC categories. Among those with *Amyoplasia*, the comorbidities were frequent and included delayed achievement of developmental milestones, seizures, obesity, and undescended testes. Comorbidities appeared to be less frequent among children with *distal arthrogryposis*, however facial dysmorphism was noted in several cases. Children included in the category *Other* were the most likely to have a comorbidity, which included a wide spectrum of issues affecting various systems (e.g., urogenital dysfunction, hernia, restrictive pulmonary syndrome) as well as specific genetic syndromes or chromosomal abnormalities (e.g., trisomy 6, Pierre Robin, multiple pterygium).

3.3. Contractures and deformities

Involvement of each joint including shoulder, elbow, wrist and hand, hip, knee, and ankle and foot across AMC classification categories are depicted in Table 1. Contractures of the ankle and deformities of the feet were very common in all three groups, affecting 91.5%

Table 2
Number of children undergoing surgical procedures per lower extremity joints and across AMC category

	Number of children who had no procedure			Number of children who had one procedure			Number of children who had more than one procedure		
	Amyoplasia (n = 47)	Distal (n = 49)	Other (n = 18)	Amyoplasia (n = 47)	Distal (n = 49)	Other (n = 18)	Amyoplasia (n = 47)	Distal (n = 49)	Other (n = 18)
At the hip level	30 (63.8%)	36 (73.5%)	10 (55.6%)	13 (27.7%)	9 (18.4%)	6 (33.3%)	4 (8.5%)	4 (8.2%)	2 (11.1%)
At the knee level	31 (66.0%)	40 (81.6%)	13 (72.2%)	11 (23.4%)	5 (10.2%)	2 (11.1%)	5 (10.6%)	4 (8.2%)	3 (16.7%)
At ankle and foot level	16 (34.0%)	18 (36.7%)	7 (38.9%)	20 (42.6%)	14 (28.6%)	6 (33.3%)	11 (23.4%)	17 (34.7%)	5 (27.8%)
At any level	12 (25.5%)	11 (22.4%)	5 (36.0%)	12 (25.5%)	12 (24.5%)	3 (16.7%)	23 (48.9%)	26 (53.1%)	10 (55.6%)

of those with Amyoplasia, 85.7% of those with distal arthrogyrosis and 83.3% of those in the *Other* category. The majority of children in all three categories had hip involvement, with 74.5% of the Amyoplasia category, 52.1% of the distal arthrogyrosis category, and 94.4% of the *Other* category. However only 15.9% of those with distal arthrogyrosis had shoulder involvement compared to 70.2% and 58.8% of those with Amyoplasia or in the *Other* category respectively. Thus, children with distal arthrogyrosis had less involvement of the proximal joints than those with Amyoplasia or those in the *Other* category. There were 16 children (14.9%) with only lower extremity (LE) involvement, including 6 with Amyoplasia, 7 with distal arthrogyrosis and 3 in the *Other* category. In addition, five children (4.3%) presented with contractures of the upper extremity (UE) only, including 2 and 3 classified as Amyoplasia and distal arthrogyrosis respectively. There were 7 children for which UE involvement was not specified. In addition, five children (4.3%) presented with contractures of the upper extremity (UE) only, including 2 and 3 classified with Amyoplasia and distal arthrogyrosis respectively. There were 7 children for which UE involvement was not specified. None of the children presented with unilateral involvement; all had bilateral involvement of at least one joint.

3.4. Orthopedic interventions

Table 2 lists the number of lower extremity surgical procedures per child across AMC category and Table 3 depicts the various surgical procedures recorded in the medical records according to joint. There were 496 surgical procedures among the 114 cases, consisting of bony surgeries, soft tissue releases, internal or external fixator and serial casting, resulting in an average of 4.35 operative procedures per participant (range: 0 to 14). The median number of surgical procedures among the participants was 4 (lower quartile = 2, upper quartile = 6). Seventeen children (14.9%) did not undergo any surgery. The most frequent site for surgical intervention

Table 3
List of surgical procedures per joint

Joint	Recorded procedures
Hip Total = 120 operations, 23 different procedures recorded	<ul style="list-style-type: none"> - Open reduction - Closed reduction - Capsulorrhaphy - Proximal femoral osteotomy - Subtrochanteric derotation osteotomy - Hip extension - Valgus derotational osteotomy - Varus derotational osteotomy - Dega osteotomy - Chiari osteotomy - Salter osteotomy - Acetabulum osteotomy (Pemberton osteotomy) - Adductor tenotomy - Hip flexor releases - Pelvic osteotomy - Hamstring release - Posterior capsule release - Pelvic support osteotomy - Arthroplasty - Distal femoral osteotomy - Femoral shortening or internal fixator - Triple osteotomy
Knee Total = 73 operations, 9 different procedures recorded	<ul style="list-style-type: none"> - Distal extension osteotomy - Supracondylar extension derotational osteotomy - Ilizarov external fixator - Soft tissue release hamstring release - Quadriceps lengthening - Epiphysiodesis (8 plates) - Posterior release - Patellar tendon reconstruction - Knee flexors release
Foot and ankle Total = 261 operations, 23 different procedures recorded	<ul style="list-style-type: none"> - Posterior, medial or posteromedial release - Achilles tendon release - Tendon transfer - Lateral Column Shortening osteotomy (salvage procedure for clubfoot) - Reduction of talonavicular joint - Vertical talus correction - Grice arthrodesis - Tarsal osteotomy - Arthrodesis - Talectomy - Ilizarov external fixator - Tibial derotation - Calcaneo-cuboid fusion or osteotomy

Table 3, continued	
Joint	Recorded procedures
	<ul style="list-style-type: none"> - Tibial derotation - Calcaneo-cuboid fusion or osteotomy - Naviculocuneiform excision/fusion/arthrodesis - Flexor digitorum longus or flexor hallucis longus tenotomy - Talar fusion - Supramalleolar osteotomy - Tibialis anterior transfer or release - Tibialis posterior lengthening or transfer - Osteochondroma excision distal tibia epiphysiodesis - Peroneus brevis transfer to tibialis anterior - Peroneus longus lengthening
Toes	<ul style="list-style-type: none"> - Flexors tendon tenotomy
Total = 11 operations, 3 different procedures recorded	<ul style="list-style-type: none"> - Fusion - Metatarsal osteotomy
Wrist	<ul style="list-style-type: none"> - Carpectomy
Total = 10 operations, 6 different procedures recorded	<ul style="list-style-type: none"> - Tendon transfer (Flexor carpi ulnaris to extensor carpi ulnaris) - Ilizarov external fixator - Fusion - Capsulotomy
Elbow	<ul style="list-style-type: none"> - Arthrolysis
Total = 8 operations, 4 different procedures recorded	<ul style="list-style-type: none"> - Triceps tendon transfer - Humeral derotation - Steindler flexorplasty
Fingers and thumb	<ul style="list-style-type: none"> - Tendon release
Total = 9 operations, 4 different procedures recorded	<ul style="list-style-type: none"> - First web space lengthening - Flexor pollicis longus lengthening - Advance ulnar collateral ligament - Metacarpophalangeal arthrodesis
Neck	<ul style="list-style-type: none"> - Sternocleidomastoid muscle release
Total = 1 operation, 1 procedure	
Spine	<ul style="list-style-type: none"> - Spinal fusion (split cord malformation release)
Total = 3 operations, 3 procedures recorded	

202 was the foot and ankle with 255 procedures within this
 203 cohort, followed by the hip with 116 procedures. Four-
 204 teen children (12.3%) underwent a surgical procedure
 205 for deformity of the upper extremity. The most common
 206 procedures performed for the UE included first web-
 207 space and thumb-in-palm releases. Ninety-two children
 208 (80.7%) had a surgical procedure to the lower limb,
 209 with the most common being foot or ankle soft tissue
 210 releases and Achilles tendon lengthening. The highest

211 number of lower extremity surgical procedures in all
 212 AMC categories (Amyoplasia, Distal and *Other*) was
 213 7. The median number of lower extremity surgical pro-
 214 cedures was 2 in the cohort (median of 1 in the Amy-
 215 oplasia group, 2 in the distal group and 2 in the *Other*
 216 group). Eleven participants (9.6%) had surgeries in both
 217 the LEs and UEs.

3.5. Functional status at latest follow-up

218
 219 The functional status of participants is described in
 220 Table 4. Among the participants for whom the goal of
 221 walking was age appropriate ($n = 44$), those with distal
 222 arthrogryposis were all community ambulators ($n =$
 223 43) expect for one participant for whom ambulation sta-
 224 tus was unspecified. Ambulation level was more varied
 225 in the Amyoplasia and *Other* groups with only 59.0%
 226 and 52.9% respectively able to ambulate, with or with-
 227 out a device. Among the children with Amyoplasia, 12
 228 of the 23 participants (52.1%) able to ambulate required
 229 a device when walking and in the *Other* group, 3 of the
 230 9 ambulatory patients (33.3%) required a device when
 231 walking. In addition, 42 participants of the whole cohort
 232 reported wearing orthoses at some point during their
 233 rehabilitation process, either knee-ankle-foot (KAFOs,
 234 $n = 11$) or ankle-foot orthoses (AFOs, $n = 31$).

235 Similarly, 97.2% of the distal arthrogryposis group
 236 were able to transfer to various surfaces in the home in-
 237 dependently while 42.3% and 71.4% of the Amyoplasia
 238 and *Other* groups respectively were autonomous during
 239 these activities. In terms of self-care, 75.0% of the group
 240 with distal arthrogryposis were independent compared
 241 to those with Amyoplasia or those in the *Other* category
 242 with rates of 28.0% and 64.3% respectively. Details on
 243 functional abilities in self-care and transfers were not
 244 specified or the subject was too young to assess these
 245 skills in about half of individuals in the Amyoplasia
 246 group.

4. Discussion

247
 248 The aim of this retrospective chart review was to de-
 249 scribe the functional status, clinical presentation, and
 250 orthopedic interventions of children with AMC who
 251 were followed in a pediatric orthopedic setting. As pre-
 252 viously described in literature [1,5], our data confirmed
 253 that AMC affects girls and boys roughly equally (52.6%
 254 vs. 47.3%). Classifying the category of AMC was done
 255 in the context of an orthopedic visit, so the use of known
 256 classification systems, such as the ones by Bamshad et

Table 4
Functional outcomes at latest follow-up depending on AMC classification

	Amyoplasia (n = 47)	Distal arthrogryposis (n = 49)	Other (n = 18)
Ambulation			
Community ambulator	11 (28.2%)	43 (97.7%)	6 (35.3%)
Ambulation with device	12 (30.8%)	0 (0.0%)	3 (17.6%)
Non-ambulator	16 (41.0%)	1 (2.3%)	8 (47.0%)
Too young/not specified	8 (17.0%)	5 (10.2%)	1 (5.6%)
Self-care			
Independent	6 (24.0%)	29 (78.4%)	9 (64.3%)
Independent with minimal assistance	9 (36.0%)	6 (16.2%)	1 (5.6%)
Dependent	10 (40.0%)	2 (5.4%)	4 (28.6%)
Too young/not specified	22 (46.8%)	12 (24.5%)	4 (22.2%)
Transfers			
Independent	11 (42.3%)	36 (97.3%)	10 (71.4%)
Independent with minimal assistance	8 (30.8%)	0 (0.0%)	0 (0.0%)
Dependent	7 (26.9%)	1 (2.7%)	4 (28.6%)
Too young/not specified	21 (44.7%)	12 (24.5%)	4 (28.6%)

al. [13] and Hall [14] was not systematic. The literature reports that over 50% of individuals with AMC are diagnosed with either Amyoplasia or distal arthrogryposis [11]. Amyoplasia is the most common form of AMC and distal arthrogryposis the second most common [15]. Our findings show a higher proportion of children with Amyoplasia and distal arthrogryposis at our Hospital than those with CNS involvement. As we are a pediatric hospital providing elective orthopedic surgery, it seems likely that children with multiple co-morbidities, particularly CNS involvement are not referred to our Hospital to the same extent as children with Amyoplasia and distal arthrogryposis.

Although several studies have identified genetic etiologies related to AMC [2,11,16–18], there is a need for identifying the genetic pathway associated with the different forms of AMC. Genetic testing is not typically offered to patients and families in community health care settings. Thus, a genetic workup should be made available to families of children with AMC to gain a better understanding of the underlying mechanisms associated with AMC, to inform them about the long-term trajectories of specific diagnoses, and for genetic counselling. Genetic testing would also help rule out other possible syndromes in the differential diagnosis that would require different management. Genetic counselling should be repositioned at the beginning of the diagnostic and therapeutic endeavor within a multidisciplinary clinical context to reach the best functional outcome possible through early, tailored therapy [19].

The majority of children with Amyoplasia (55.5%) had involvement of all three joints of the UEs, which might explain the finding that only 28.0% were independent walkers, and that 32.0% required technical or human aid to achieve independence in self-care activ-

ities. In the distal group, joint involvement is much less predominant at the levels of the shoulder and elbow and not surprisingly, rates of autonomy in self-care are much higher (e.g., 75.0% versus 28.0%). This finding suggests that proximal upper extremity involvement is a critical factor in reaching independence in self-care. Nevertheless, only a small percentage of the children with Amyoplasia had UE surgery to address these contractures. The paucity of scientific literature on the effectiveness of interventions in the UE in AMC highlights the need for research on operative and non-operative outcomes in this area. It has been suggested in the literature that coordination of upper and lower limb surgical procedures can be a good strategy to limit anesthetic exposure and maximize rehabilitation early in life [20]. The annotated definition of AMC proposed by Dahan-Oliel and colleagues in 2019 states that spinal deformities may be present at birth or may develop during childhood and adolescence [15]. Although we did not report on spinal involvement for the 114 children in this retrospective chart review, three children or 2.6% of the cohort had spinal fusion. Given the broad age range of our sample, it is possible that other participants eventually required surgical treatment of their spinal deformities at a later date, which was not captured by our review. As our Hospital provides elective orthopedic care, some spinal surgeries for children with intubation difficulties are performed at an affiliated children's hospital that has an intensive care unit. Therefore, some of the cases reviewed may have had spine surgeries that were not reported at our Hospital.

Children in all three classification groups had joint involvement of all three levels of the lower extremity. Not surprisingly, those with distal AMC were found to have the least hip involvement, 52.1%. The Amyoplasia

group were most affected at the foot and ankle, with 91.5% of the participants being recorded with unilateral or bilateral contractures at that level. Relatively less involvement of the hip and knee appears to be associated with independence in ambulation and transfers as noted by the high rates of community ambulation without devices in those with distal arthrogyposis. In contrast to the UE, interventions addressing the LE issues were frequent. This finding correlates with the incidence of 5 subjects (4.4%) in our review with only UE involvement. Children with joint contractures of only the UE or LE may not be readily detected with AMC, as contractures of several different body parts may be associated with a more characteristic AMC phenotype. Studies have also shown that even with severe initial presentation, children with AMC can achieve functional outcomes through early treatment interventions and individualized and aggressive management [21–24]. Documentation regarding rehabilitation as well as the use of standardized functional measures was lacking in the charts reviewed, leading us to infer that referral to rehabilitation, particularly in the early years of this review was inconsistent. Functional outcome measures inform evidence-based practice and are essential for the evaluation of treatment effectiveness. In lieu of the use of outcome measures, a classification system to describe ambulation was used which was based on previous work of Dr. van Bosse (personal communication). This classification system was then extrapolated for describing self-care and transfers. This simple classification system enabled accurate description in a large sample of patients in AMC despite the retrospective nature of this study.

Our findings of multiple joint involvement, the extensive orthopedic management as well as the need for aids in daily activities are in line with the current literature [25–27] and emphasize the importance of early referral to rehabilitation and orthopedics. Therefore, it is suggested that the management of children with AMC should be within a multidisciplinary context, and should encompass genetics, orthopaedics, nursing, rehabilitation, social services, orthotics, and other professions to meet the spectrum of needs children with AMC may experience. This is in line with previous studies that demonstrate the importance of integrating the care of children with AMC with a multidisciplinary team approach [19,28]. Indeed, a multidisciplinary approach could potentially diminish waiting times for families, provide specialty coordinated care across several health care disciplines in one setting, ensure early genetic evaluation, and improve the quality of care provided as has

been demonstrated in the care of children with chronic musculoskeletal conditions [29,30]. For these reasons, we established a monthly multidisciplinary clinic for children with AMC in 2016, which provides coordinated care from the following disciplines: orthopedic subspecialties in upper extremities, lower extremities and spine, rehabilitation, social work, radiology, dietetics, orthotics, and metabolic investigation [31]. As our Hospital provides care to children with musculoskeletal conditions across the geographic landscape, our team piloted the remote delivery of a home exercise program for youth with AMC living across Canada [32]. The pre-established clinic model and prior use of telemedicine in AMC greatly facilitated the provision of continued care to our patients when the COVID-19 pandemic manifested in March 2020. Our experience pre-COVID-19 helped us immensely in facing the realities of a pandemic while providing care, and demonstrated that our clinic model could also be delivered utilizing a remote approach. Indeed, telemedicine plays a key role in prioritizing the treatment of common orthopaedic conditions and is a safe modality to deliver care when physical distancing measures must be implemented [33]. In fact, in July 2020 with traveling restrictions still in place, the multidisciplinary AMC clinic was delivered via telemedicine. The use of telemedicine to deliver multidisciplinary care has also been implemented for other populations with rare musculoskeletal conditions, including osteogenesis imperfecta, and could be applied to other pediatric populations at our center, such as skeletal dysplasias and neuromuscular conditions.

Limitations of this retrospective review include the lack of routine genetic testing which precluded the provision of a specific AMC diagnosis. Due to the lack of information on the underlying genetic diagnosis, categories of AMC may have been misclassified. Furthermore, the inconsistent referral of patients to rehabilitation services resulted in limited information of functional status. In cases when patients were referred to rehabilitation, a standardized evaluation protocol was missing. Therefore, the evaluation of the effectiveness of specific interventions was not possible. This highlights the need for the development of guidelines for early diagnosis and management of arthrogyposis as well as long-term follow-up using standardized functional outcomes. As children with arthrogyposis have a heterogenous presentation, the development of clinical tools specific to their wide range of physical presentations should be a research priority.

As AMC is rare, the relatively large sample size of this chart review allowed better insight and understand-

ing into the challenges associated with multiple congenital contractures. Severe upper limb involvement is clearly associated with more challenges to reaching autonomy in self-care. Lower limb involvement at the proximal level is often associated with a poorer prognosis in terms of mobility outcome. However, this also highlights that children with distal arthrogryposis can usually be independent in self-care, transfers and mobility and should be treated with those objectives in mind.

Moving forward, multi-site prospective studies are required in the fields of genetics, treatment effectiveness and adult outcomes [34]. The findings of this study demonstrate the importance of genetic testing to provide accurate diagnosis and classification, researching and promoting the use of standardized functional measures in AMC, and the provision of care within a multidisciplinary context.

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Conflict of interest

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