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# Muscle and joint function in children living with arthrogryposis multiplex congenita: A scoping review

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**Abstract**

Arthrogryposis multiplex congenita (AMC) is characterized by congenital joint contractures present in two or more body areas. Lack of fetal movement is the underlying cause of AMC, which can lead to abnormal connective tissue surrounding the joint resulting in stiffness and muscle atrophy. Treatment aims at improving function and mobility through surgical and/or conservative interventions. A scoping review was conducted to explore the existing knowledge of the evaluation and treatment of muscle and joint function in children with AMC. Three search engines were included and identified 1271 articles. Eighty-seven studies met the selection criteria and were included in this review. All included studies focused on joints, 30 of which also assessed the muscle. Assessment most often included the position of the contractures (n=72), as well as range of motion (n=66). Interventions to improve muscle and joint function were reported in 82 of the 87 papers and included surgery (n=70) and conservative interventions (n=74) with bony surgery (i.e. osteotomy) the most common surgery and rehabilitation the most common conservative intervention. Recurrences of contractures were mentioned in 46 of the 68 studies providing a follow-up. Future studies should use validated measures to assess muscle and joint function, and conservative interventions should be described in greater detail and to include a longer follow-up.

**Key Words:** arthrogryposis multiplex congenita, pediatric, muscle, joint, management

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## 1. INTRODUCTION

Arthrogryposis affects 1 in 3000 to 1 in 4300 live births (Lowry, Sibbald, Bedard, & Hall, 2010). Arthrogryposis multiplex congenita (AMC) and arthrogryposis are interchangeable terms used to describe the presence of multiple congenital contractures affecting at least two different areas of the body. A contracture is defined as “the limitation of movement of a specific joint”, in other words, a joint that does not have a full range of movement (Staheli, Hall, Jaffe, & Paholke, 1999). While these terms may have been used as a diagnosis in the past, the multiple and varying etiologies that result in AMC have led to their recognition as descriptive signs (Bamshad, Van Heest, & Pleasure, 2009; Hall, 2014). The contractures associated with AMC are generally non-progressive and can affect the upper limbs, lower limbs, spine and jaw, which may result in limited independence in self-care and mobility, and in restricted participation in daily life (Amor, Spaeth, Chafey, & Gogola, 2011; Dillon, Bjornson, Jaffe, Hall, & Song, 2009; Joubert & Franzsen, 2016).

While different etiologies are responsible for the development of arthrogryposis, all are associated with fetal akinesia. Abnormalities in the central or peripheral nervous system can lead to diminished movement in utero. Genetic mutations and myopathic processes can lead to developmental abnormalities in muscles resulting in fetal immobility (Staheli et al., 1999). Additionally, a variety of other factors such as maternal illness, space limitation, intrauterine reduce blood supply or drug use can all lead to diminished mobility during gestation (Hall, 2014). When fetal akinesia occurs, the lack of joint movement can lead to abnormal connective tissue surrounding the joint resulting in stiffness (Ferguson & Wainwright, 2013). It can also result in further disuse of the muscle mobilizing the joint, impacting its development and leading to fibrosis of the muscles and tendons leading to a muscle weakness. All of these factors contribute to the formation of contractures (Ferguson & Wainwright, 2013; Hall, 2014). Current clinical treatments to overcome these contractures are based on a conservative approach (i.e. non-surgical) as well as surgical management, both aiming at improving function and mobility (Oishi et al., 2017; H. J. P. van Bosse et al., 2017).

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The World Health Organization recognizes that function is dependent on the interplay between various intrinsic and extrinsic factors. The International Classification of Functioning, Disability and Health (ICF) emphasizes the interaction between an individual's body structures and functions, activity, participation, personal and environmental factors, to optimize function and minimize disability (W.H.O, 2002). Recently, in a study published by Elfassy et al. (2019) youth reported that their ability to participate in meaningful activities and roles are affected by their environment, such as the home, school and community, and is further impacted during transition periods. Moreover, clinicians reported a need to develop an AMC-specific outcome tool and best practice guidelines. According to Rumrill, Fitzgerald, and Merchant (2010), scoping reviews of the literature is an efficient way of identifying themes and trends in high-volume areas of scientific inquiry. They are used to review and to synthesize existing literature. The aim of this scoping review was to identify how muscle and joint function are evaluated and treated among children with AMC.

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## 2. MATERIALS AND METHODS

A scoping review was chosen to better comprehend the existing knowledge on the evaluation and treatment of muscle and joint function in children with AMC. Arksey and O'Malley (2005) framework with the additional recommendations by Levac, Colquhoun, and O'Brien (2010) was used as the methodological approach to obtain the best synthesis of our findings. The Preferred Reporting Items for Systematic Reviews and Meta-Analyses extension for Scoping Reviews (PRISMA-ScR) checklist was used (Tricco et al., 2018).

### Search Strategy and Selection Criteria

A search of the literature was conducted using three bibliographic databases: Medline (1946-June 2018), PsycInfo (1987-June 2018) and CINAHL (1955-June 2018). The search terms included a combination of terms related to arthrogryposis multiplex congenita ("AMC", "arthrogryposis", "amyoplasia" and "multiple congenital contractures") with those related to muscle and joint function ("muscle", "function," "joint," "strengthening," "training," "exercise" and "endurance") combined using "AND" or “OR.” The search strategy was developed for Medline and was then adapted for other search engines. All identified studies were exported into EndNote for removal of duplicates. The remaining studies were exported into Rayyan, a free systematic review web application which allows for online screening. Two research assistants screened titles, abstracts and full texts of all identified studies through a blinded review (Ouzzani, Hammady, Fedorowicz, & Elmagarmid, 2016). In the case of a disagreement, a third member of the research team (N.D-O) was consulted to make a decision regarding inclusion or exclusion. Refer to table 1 for the selection criteria.

Data Extraction and Synthesis

To identify study characteristics and important findings on muscle and joint function, a data extraction sheet in Excel was created. The general information (country, date of publication, study type, objectives), methods (population characteristic, intervention, evaluation), results, and conclusions of the included studies were extracted. Two authors of the manuscript (M.G. and K.C.) independently extracted the data of the first ten research papers with the data extraction sheet. Once completed, they compared extracted data, discussed discrepancies and adjusted the data extraction form to ensure minimal data extraction discrepancies. After the validation of the data extraction form, remaining articles were divided between them for extraction. They met every 10–15 articles were extracted to ensure the data extraction process remained in line with the research question. Once the data were extracted from all the included studies, findings were synthesized.

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## 3. RESULTS

Search Strategy and Study Selection

The search strategy yielded a total of 1271 articles. Following removal of duplicates, 1077 titles and abstracts were screened, and 136 full-text articles reviewed for eligibility. Eighty-seven studies met the selection criteria and were included in this scoping review. Refer to Figure 1 for the PRISMA flow chart.

Study Characteristics

Of the 87 publications retained, 22 different countries were represented, with three studies yielding from a collaboration between two countries. The United States published the largest number of studies (n=39), followed by Japan (n=6) and Canada (n=5). The oldest publication was in 1955. There were 32 publications between January 2010 and June 2018. With six publications, 2013 was the year with the largest number of publications on the topic. Study design was mainly retrospective, refer to figure 2 for other study designs represented. The level of evidence was not mentioned in 71 of the 87 papers and the remaining was classified as level II (n=2), level III (n=1), level IV (n=12) and level V (n=1). Seven of the 87 studies reported having received funding.

<u>Sample Characteristics</u>

The 87 included studies result in a total of 1426 participants (425 females, 464 males, 537 undefined, age range= birth to 21 years). Age at interventions ranged from under 12 months (n=19) to greater than 60 months (n=25), with six studies providing an intervention at 12-24 months, seven studies at 24-36 months, three studies at 36-48 months and nine studies at 48-60 months; time at intervention was not reported in nine studies. Follow-up period post-intervention ranged from under 12 months (n=11) to greater than 48 months (n=33), with ten studies providing a follow-up period of 12-24 months and nine studies with 24-36 months; a follow-up period was not reported in six studies. Of the 87 studies, 34 reported on type of AMC, which Amyoplasia most commonly studied. Upper extremity (i.e., hand, elbow and shoulder) and lower extremity (i.e., hip, knee and foot) were studied almost equally with 43 and 48 articles respectively.

<u>Assessment and management of joint and muscle function</u>

A variety of different assessments, tools, methods and grading scales have been used to assess muscle and joint function in children with AMC depending on the body part examined (Tables 2-4). A homemade grading scale was used in 12 studies. All included studies (n=87) focused on joints, 30 of which also assessed the muscle. Contractures' position, range of motion (ROM), strength evaluations and bony/muscle composition were assessed for each joint. ROM was measured actively (n=14), passively (n=12), or both (n=19). Description of the arc of motion was present in 23 studies.



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Muscle strength was most commonly assessed using the 5-point Medical Research Council Grade generally or generally reported as manual muscle testing (MMT) (n=18). Muscle signal transmission was measured by electromyography (EMG). Muscle endurance was not assessed in any study.

Other considerations related to the muscle included muscle histology, level of creatine kinase and electron microscopy. The level of creatine kinase was found to be minimally elevated in one of the eight patients tested and electron microscopy showed in one of the 10 patients tested subtle changes with irregular myofribrils (Adams, Becker, & Murphy, 1988). They also found abnormal muscle fibers histology in nine of 13 patients, including abnormal predominance of type I fibers in five children over 10 months of age, and type II fibers in 3 children between 2 to 10 weeks of age. As well, two children had an atrophy of type II muscle fibers. Nerve testing was another aspect assessed in two studies. Bahm (2013) reported having used intraoperative electrical stimulation testing during their exploratory surgery. Hall, Reed, and Driscoll (1983) did a CNS microhistopathology and a biopsy of a peripheral nerve but they found no apparent changes.

Quantified gait analysis using an optoelectronic camera system was done to assess gait deviations from norms and efficiency of lower limb orthoses in correcting gait or posture (Bartonek, Eriksson, & Gutierrez-Farewik, 2007; Bartonek et al., 2011; Bohm, Dussa, Multerer, & Doderlein, 2013; Eriksson, Bartonek, Ponten, & Gutierrez-Farewik, 2015; Haumont et al., 2011). Eriksson et al. (2015) found that all AMC groups (i.e., wearing KAFO, AFO, FO or no orthoses) showed less hip extension than the control group, but hip flexion moment was significantly lower only in children wearing KAFO, which can be attributed to their gait strategy with bilateral locked KAFOs. Children, who had weak knee extensors, were helped by their locked KAFOs and therefore showed similar knee extension moment as the other groups. This study demonstrated the children's high reliance on hip muscles and presumably trunk muscles to provide propulsion and showed that with adequate orthotic support, children with AMC and even with severe weakness and contractures can achieve walking. Bohm et al. (2013) found that AMC patients' gait showed excessive range in thorax obliquity in 10 of the 18



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children and suggested that excessive thorax obliquity during gait could be associated with reduced strength and mobility of the hip. Bartonek et al. (2007) found ankle absorption at foot strike was abnormally low in AMC patients and that carbon fiber spring orthoses were more efficient at increasing ankle absorption at foot strike then regular orthoses (KAFO or AFO). Bartonek et al. (2011) assessed the influence of different heel lifts condition (i.e., 0˚, 10˚, 20˚, 30˚) and between orthoses and non-orthoses users on static posture. Significant changes were seen between various heel lifts in ankle, knee and pelvis in AMC when it was only seen in the ankle for the control group. Between orthoses and non-orthoses users, significant differences were seen between different heel lift conditions: the ankle angle indicated that higher heel lifts lead to more plantarflexion (non-orthoses); the knee angle increased in flexion with higher heels lifts (Orthoses); the pelvic angle change indicated that increasing heel lift decrease anterior tilt (Orthoses and non-orthoses). Apply heel heights adequate to each individual's orthopaedic and neurologic conditions to improve biomechanical alignment with respect to all body segments is a challenge.

Interventions to improve muscle and joint function were reported in 82 of the 87 studies (94%) and seemed to occur mostly within the first year of the child's life (19/82, 23.2%) or after the age of five (25/82, 30.5%). Interventions were surgical in 70 studies (85%) and conservative in 74 studies (90%). Surgical management included bony surgery, soft tissue surgery, transfers of muscle, tendon and nerve, muscle and tendon lengthening, and joint reduction (Table 5). Conservative management included splinting, bracing, casting, orthoses, harness and rehabilitation (Table 6). Physical and occupational therapy, kinesitherapy, and home exercise programs were considered as rehabilitation. Conservative procedures were considered either as the main intervention or as an adjunct to surgery. Of the 74 papers that included conservative treatments, 31% (23/74) mentioned this type of intervention but did not provide details on the frequency, duration and treatment modality. In the other 69% of studies reporting on conservative treatment, details on the intervention were minimal in most cases and focused mainly on reporting casting or splinting duration of treatment. Among the 44 studies reporting on rehabilitation, only 27% (12/44) provided the description and/or duration of treatment. Results are presented according to the joint affected in the following sections.



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*Head and Jaw (n=6)*

Two studies provided an assessment of the head and four focused exclusively on the jaw. Of those, Azbell and Dannemiller (2015) measured the passive range of motion of the lateral cervical flexion and the cervical rotation of an infant and found limited cervical rotation (R 90˚, L 70˚) and lateral cervical flexion (R 40˚, L10˚) at 11 days of age. Muscle tone of the head was assessed in one study, with a finding of reduced head muscle tone at birth (Binkiewicz-Glinska et al., 2013). Three of the four studies on the jaw were single-case studies and one included 23 patients with five having jaw contractures. The jaw was measured using different methods. Skeletal open bite was used in two studies that reported values of 15 and 16 mm (Heffez, Doku, & O'Donnell, 1985; Kargel, Dimas, & Chang, 2007), and another study reported mild anterior open bite, but without providing specific values (Epstein & Wittenberg, 1987). Mouth opening was also assessed using interincisal opening (12 mm) (Epstein & Wittenberg, 1987) or maximal incisal opening (25 mm) (Kargel et al., 2007). Those limited mouth opening was reported to affect oral hygiene, speech (Epstein & Wittenberg, 1987) and feeding (Kargel et al., 2007). Steinberg, Nelson, Feinberg, and Calhoun (1996) mentioned feeding issues secondary to weakened masticatory musculature. In addition, muscle activity was assessed with EMG in two studies. Heffez et al. (1985) found hyperactivity of the suprahyoid muscle group and hypoactivity of anterior and middle temporalis and external pterygoid muscles. Epstein and Wittenberg (1987) reported EMG results of the temporalis and masseter muscles showing normal spontaneous evoked activity. Bone imaging tools were common as radiography and tomography were used by Epstein and Wittenberg (1987) and Heffez et al. (1985), and CT scan were used by Steinberg et al. (1996). Following radiographs and joint tomograms, two studies found the absence of translational movement of the condyles (Epstein & Wittenberg, 1987; Heffez et al., 1985).

Interventions to improve ROM of the head or jaw were reported in four of the six studies. Azbell and Dannemiller (2015) mentioned 35˚ of improvement of the left lateral cervical flexion (10˚ to 45˚) after a nine-month PT and OT intervention. Epstein and Wittenberg (1987) reported several surgical procedures including an intraoral



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coronoidectomy, a temporo-mandibular arthroplasty, a condylectomy and a temporal eminence reduction followed by a home exercise program in one patient. Following this treatment regimen, the patient had an interincisal opening improvement of 11 mm at 16-month follow-up. Kargel et al. (2007) increased the maximal incisal opening by 6 mm after skeletal osteotomies and repositioning of the jaw as well as an orthodontic treatment. PT including stretching exercises was reported in two studies (Epstein & Wittenberg, 1987; Steinberg et al., 1996). Exercises in two patients with limited jaw opening provided gains that were not maintained following therapy (Steinberg et al., 1996).

*Shoulder (n=13)*

Shoulder position was reported to be in internal rotation (Hall et al., 1983; Kroksmark, Kimber, Jerre, Beckung, & Tulinius, 2006) and one case in elevated and forward position (Bharucha, Pandya, and Dastur (1972). Studies using ROM measured flexion (0˚ to 120˚), extension (0˚ to 20˚), abduction (40˚ to 180˚), internal rotation (excessive to 90˚) and external rotation (0˚ to 70˚) (Babik et al., 2016; Kroksmark et al., 2006; Sala, Rosenthal, & Grant, 1996). Shoulder strength was assessed in two studies. Bharucha et al. (1972) found by EMG investigation a weakness of the shoulder muscles in nine cases out of 16 but with different distribution. In one case, deltoid muscles were thin and in some other cases the entire shoulder girdle musculature was weak. One case showed a weakness in all muscles supplied by C5 and C6 nerve roots. Using a hand-held dynamometer, Kroksmark et al. (2006) found diminished shoulder abductor strength in the non-ambulatory group compared to the community ambulatory group.

Surgical correction of the shoulder was minimal and included two studies using bony surgery to correct an internal rotational deformity. Bennett, Hansen, Granberry, and Cain (1985) did a derotation osteotomy and achieved good functional results to position hand in front of the body. Hahn (1985) did 12 proximal humeral osteotomies and they suggested doing this kind of surgery on older children. However, they did not mention specific results for this surgery.



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Rehabilitation aimed to increase shoulder ROM and lasted between 11 and 36 weeks (Azbell & Dannemiller, 2015; Sala et al., 1996). All three studies showed an improvement of 25˚ to 80˚ in shoulder flexion (Azbell & Dannemiller, 2015; Binkiewicz-Glinska et al., 2013; Sala et al., 1996). For shoulder abduction, 0˚ to 40˚ of improvement was found (Azbell & Dannemiller, 2015; Sala et al., 1996). No improvement was noted for internal and external rotation (Azbell & Dannemiller, 2015; Sala et al., 1996). Babik et al. (2016) was the only study to use a 3-month intervention using an exoskeleton to look at the effect on the shoulder on an 8-month-old infant. They found an improvement of active ROM from 15˚ to 115˚ depending on the position (sitting or supine) and the side evaluated.

*Elbow (n=31)*

Passive ROM varied from no restriction to complete restriction of movement (0˚ to full ROM) (Ramirez, Richards, Kozin, & Zlotolow, 2017; Zargarbashi, Nabian, Werthel, & Valenti, 2017). Extension contractures were most common in general (n=13), while a greater proportion of participants with elbow flexion contractures was reported in three studies (Ayadi et al., 2015; Hahn, 1985; Kite, 1955). Radiography was used in two studies and showed elbow dislocations (Bharucha et al., 1972; Sala et al., 1996). All papers assessing muscle strength before elbow surgery reported no bicep strength except two who found a bicep strength of 2-3/5 (A. Van Heest, James, Lewica, & Anderson, 2008; Williams, 1973; Zargarbashi et al., 2017). Four studies reported complete or severe atrophy of the biceps and brachialis muscles using electromyography (EMG) (Bharucha et al., 1972; Carroll & Hill, 1970; Chomiak, Dungl, & Vcelak, 2014; Doi, Arakawa, Hattori, & Baliarsing, 2011), while one study reported an EMG signal in those muscles with no associated active movement (Chomiak et al., 2014). A dynamometer was used in two studies to assess elbow muscle strength allowing more objective values (Doi et al., 2011; Kroksmark et al., 2006).

In terms of elbow surgery, pectoralis major, triceps, latissimus dorsi, deltoid and gracilis muscles were transferred to increase active flexion ROM and muscle strength (Atkins, Bell, & Sharrard, 1985). After a combination of tricepsplasty and tricep transfer and an average follow-up of almost 5 years among 23 elbows, muscle strength ranged



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from 0 to 5 on a scale of 5 (Williams, 1973). Assessing the capacity to lift a weight, Goldfarb, Burke, Strecker, and Manske (2004) found that after a Steindler Flexorplasty surgery, all patients had a flexion strength of at least 1⁄4 kg through their entire arc of flexion with a maximum flexion strength of 4 kg compared to any active flexion of the elbow against gravity prior surgery. Lahoti and Bell (2005) mentioned that early results in all patients were encouraging. However, after a long follow-up of 7 to 19 years, gradual and progressive increase in flexion deformity in eight of the ten elbows was observed without any change in the power of the muscle transferred. The only patient who retained good long-term function had surgery at a later age of 14 years. Repair of innervation assessed by EMG was found following pectoralis major muscles transfer during a follow-up period of 13–16 years (Chomiak et al., 2014). Nerve transfer surgery found active elbow flexion at six months post-surgery (Bahm, 2013). Soft tissue release, such as capsulotomy, was often combined with triceps lengthening to increase elbow passive range of motion to 70–90˚ when conservative treatment did not work, followed by muscle/tendon transfer to obtain active ROM (Chomiak et al., 2014; Goldfarb et al., 2004). After a combination of triceps lengthening and capsulotomy, A. Van Heest et al. (2008) reported an improvement in the arc of passive motion of all twenty-nine elbows from an average of 32˚ (range, 0˚ to 75˚) preoperatively to an average of 66˚ (range, 10˚ to 125˚) postoperatively.

Conservative treatment was done before (n=9) and after surgery (n=13) or as main treatment (n=6) with the goal to increase passive ROM to 70˚ or 90˚ preoperatively (Doi et al., 2011; Goldfarb et al., 2004). Serial casting or splinting was reported in two studies (Fassier, Wicart, Dubousset, & Seringe, 2009; Goldfarb et al., 2004). Studies reported improved passive ROM following rehabilitation that did not require surgery (Binkiewicz-Glinska et al., 2013; Doi et al., 2011; Martin, Perrot, & Duteille, 2009). Other studies reported that rehabilitation did not provide sufficient passive ROM for transfer surgery eligibility, thus requiring soft tissue release or lengthening to obtain the required passive ROM for a muscle transfer. Goal of rehabilitation post-surgery is indicated to reduce recurrences of contractures. Casts were kept for a period ranging from three to six weeks in a flexion position (Atkins et al., 1985; Gagnon, Fogelson, & Seyfer, 2000). Splints were used for a period ranging from four weeks to six months (Goldfarb et al., 2004;



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Mennen, 1993). Post-surgery rehabilitation included muscle re-education and passive and active mobilisation. Doi et al. (2011) used electromyography biofeedback exercises to strengthen the transferred muscles. Results following strictly conservative treatment consisting of splinting and rehabilitation (i.e., stretching and strengthening) were reported to last between 15 weeks and 12 months and started early (i.e., 7 days-4 weeks of age) and to increase passive ROM (20˚ - 60˚) in all studies (Azbell & Dannemiller, 2015; Kamil & Correia, 1990; Sala et al., 1996). The use of an exoskeleton was found to improve daily activities such as feeding (Haumont et al., 2011) and assisted in elbow flexion (Babik et al., 2016).

*Hand (n=26)*

This section includes studies investigating the hand, thumbs, fingers and wrists. Wrist contractures were mostly in flexion with ulnar deviation except for two studies that reported wrist extension contractures in few patients (Kite, 1955; Mead, Lithgow, & Sweeney, 1958). Thumb deformities included thumb in adduction (i.e., clasped thumb deformity) (Abdel-Ghani, Mahmoud, Shaheen, & Abdel-Wahed, 2017). Radiography was used in six studies to assess the position of the wrist, thumb and finger, and was used by Abdel-Ghani et al. (2017) to assess bony structures and found ossification of the epiphysis of the proximal phalanx of the thumb. ROM at the wrist ranged between 0˚ to 42˚ (Call & Strickland, 1981; Wenner & Saperia, 1987) and 0˚ to 30˚ for thumb abduction (Abdel-Ghani et al., 2017). Muscle strength or function was assessed most of the time using pinch grasp (Bharucha et al., 1972; Dangles & Bilos, 1981) or grip strength measured with a dynamometer (Kroksmark et al., 2006; Wenner & Saperia, 1987). Kroksmark et al. (2006) assessed wrist extensor strength and found lower values among non-ambulators compared to community ambulators. Weakness of the intrinsic hand muscles of a patient was reported by Call and Strickland (1981) with no mention of how it was assessed.

Soft-tissue surgery included volar wrist release, z-plasty and web space deepening. Of the outcomes of 15 wrist procedures reported by Bennett et al. (1985), two that had undergone only soft tissue release failed. Opponensplasty was the transfer procedure most frequently reported for the thumb (Dangles & Bilos, 1981; Takagi, Seki,

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Takayama, & Mochida, 2016). Wrist transfers included flexor carpi ulnaris and extensor carpi ulnaris (Bennett et al., 1985; Ezaki & Carter, 2004). Chondrodesis of the thumb metacarpophalangeal joint, carpectomy, ulnar osteotomy and wrist fusion were some of the bony surgery performed. Of the four studies including a lengthening procedure, three mentioned lengthening the flexor pollicis longus tendon, and one study lengthened the flexor pollicis profondus tendon (Call & Strickland, 1981) resulting in a better functional position without improvement in ROM (Ezaki & Carter, 2004; Foy, Mills, Wheeler, Ezaki, & Oishi, 2013).

Rehabilitation and splints were used before surgery but were unsuccessful at treating digital contractures or excessive wrist contractures. (Call & Strickland, 1981; Foy et al., 2013; A. E. Van Heest & Rodriguez, 2013). Post-surgery, cast were used most of the time until bone consolidation (4 to 9 weeks) (Ezaki & Carter, 2004; Tsuyuguchi, Masada, Kawabata, Kawai, & Ono, 1985). Splints were kept for 3 weeks to 6 months (Call & Strickland, 1981; Mennen, 1993). For studies using rehabilitation as main course of treatment, Smith and Drennan (2002) used a serial casting procedure among patients with classic and distal arthrogryposis. An average of four casting sessions per patient (mean length = 56 days per session) led to better results for the distal arthrogryposis group than the classic group. With regard to rehabilitation, Sala et al. (1996) provided an early intensive rehabilitative and splinting treatment from four weeks to 15 weeks of age on a patient. The patient received PT and OT treatments between twice daily to 2-3x/week and wore a wrist splint for 16h/day. An improvement of wrist extension (40˚ - 45˚) and active flexion of her right wrist and digits was reported at the final assessment.

*Spine (n=11)*

Spine deformities were diagnosed from birth to 13 years and included scoliosis (n=8) and lordosis (n=4) (Fassier et al., 2009; Herron, Westin, & Dawson, 1978). For studies assessing spine deformities through radiography, an initial thoracolumbar deformity ranging 16˚ to 108˚ was found (Herron et al., 1978; Imagama et al., 2013; Sala et al., 1996). Lordosis and kyphosis were also considered in Herron et al. (1978) and Imagama et al. (2013). CT scan and 3D-CT showed a compression of the right bronchus block by an intrathoracic vertebral protrusion and an atelectasis of the right inferior lobe



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with a vital capacity of 0.33L, indicative of severe restrictive ventilator impairment in one 7-years old girl (Imagama et al., 2013). A good spine alignment is important for sitting, walking but also for swallowing and for respiratory function (Fassier et al., 2009; Imagama et al., 2013; Sala et al., 1996).

Three studies provided spinal surgery. Imagama et al. (2013) did an anterior and posterior bony fusion on a 7-year old girl to correct a scoliosis from 65˚ to 14˚ and 76˚ to 42˚ for the thoracolumbar lordosis without loss of correction three years after surgery. Moreover, atelectasis of the right lobe was resolved, vital capacity improved to 0.53L and posture was improved leading to a stable gait. Herron et al. (1978) did 10 hip surgery to correct pelvic obliquity with the aim to control the spinal deformity, which worked for two patients. Spinal fusions were performed in the seven remaining patients, and post-operative complications occurred in five of those patients (e.g., pseudoarthrosis combined with loss of correction, curve progression either inferior or superior to the fusion and worst curves after surgery). The two patients having the worst pre-operative curve (75˚ and 108˚) underwent an osteotomy combined with a spinal fusion and had better post-operative results with an improvement of 30˚ and 24˚, but the scoliosis was not completely resolved. Fassier et al. (2009) did a spinal fusion on two patients (14 and 19 years) but did not give any results.

Bracing was used as primary conservative treatment in four studies, but spinal fusion was required in three of these studies due to a curve progression (Fassier et al., 2009; Hahn, 1985; Herron et al., 1978; Imagama et al., 2013). Herron et al. (1978) mentioned that at best, braces only reduced or delayed the progression of the curve and permitted the maturation of the spine to perform spine fusion more effectively. Fassier et al. (2009) treated four patients with a brace at a mean age of 8.6 years of whom two needed a spinal fusion. Casting gave similar results to bracing in the study of Herron et al. (1978) and Hahn (1985) did not mention results of casting. Traction was used in two studies; specific results following traction were not provided (Hahn, 1985; Herron et al., 1978). Hahn (1985) used manual trunk stretching every two hours for the first few weeks of life in five infants, combined with the use of a harness to maintain three-point force. Sala et al. (1996) combined stretching to the trunk musculature and rehabilitation, followed by the use of a supine positioner and spine orthosis over a period of 9 weeks to achieve neutral alignment in a 4-week-old patient. At discharge, trunk alignment in supine was improved, and improved active head rotation.

*Hip (n=27)*

Hip flexion contractures were often accompanied by abduction and external rotation, although three cases of hip extension contractures were reported (Bharucha et al., 1972; Fassier et al., 2009; Mead et al., 1958). Radiography allows to assess hip position with specific measures, such as the acetabular index (28˚ to 50˚ before surgery) and the centre-edge angle (Asif, Umer, Beg, & Umar, 2004; Aydin, Yilmaz, Senaran, & Durgut, 2016; Rocha, Nishimori, Figueiredo, Grimm, & Cunha, 2010; Szoke, Staheli, Jaffe, & Hall, 1996). Using ultrasound to assess initial hip position, Binkiewicz-Glinska et al. (2013) reported a good seating of the femoral head of the acetabulum and using Graf's classification, Aydin et al. (2016) noticed that all hips were classified as type 4 (i.e., dislocated). Initial ROM of the hip varied; flexion (40–140˚), extension (-70-15˚), abduction (5–65˚), adduction (-5–50˚), internal rotation (-20–60˚) and external rotation (5–90˚) (Aydin et al., 2016; Azbell & Dannemiller, 2015; Bohm et al., 2013; Kroksmark et al., 2006). Muscle strength was tested in flexion (MMT=1–5), extension (MMT=1–5) and abduction (MMT=2–5) (Bartonek et al., 2007; Bohm et al., 2013; Eriksson et al., 2015; Moore, Major, Stallard, & Butler, 1990). Hip adduction strength (MMT=2—5) (Bartonek et al., 2007; Moore et al., 1990), hip internal rotation (MMT=2/5) and external rotation (MMT=5/5) (Moore et al., 1990) were measured. Kroksmark et al. (2006) measured hip isometric muscle strength with a hand-held dynamometer allowing to compare muscle strength in children with different ambulation levels. Non-ambulators and house ambulators had significantly weaker hip flexors than community ambulators ($p < 0.001$), hip extensors ($p < 0.001$) and hip abductors ($p < 0.001$ and $p = 0.001$).

Surgical correction of the hip was done between 3 months and 14 years of age. Hip reduction was performed to correct dislocation; six studies used closed reduction initially but either failed to resolve the dislocation or results were not provided(Akazawa et al., 1998; Fassier et al., 2009; Hahn, 1985; Sud, Kumar, & Mehtani, 2013; Wada et al., 2012; Yau, Chow, Li, & Leong, 2002). Yau et al. (2002) was the only study to mention



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successful closed reduction on four hips that were subluxed following correction for dislocation. Open reduction to correct hip dislocation or subluxation (n=14 studies) led to improvements of the acetabular index ranging from 13.7˚ to 25˚ as reported in three studies (Asif et al., 2004; Rocha et al., 2010; Szoke et al., 1996). Asif et al. (2004) reported an improvement of the centre-edge angle from -41˚ pre-operatively to 18˚ at a mean follow-up of four years. Complications included hip re-dislocation or subluxation, and avascular necrosis (Aydin et al., 2016; Fassier et al., 2009; Rocha et al., 2010; Szoke et al., 1996). Even if hips were successfully reduced, functional results were not always optimal (Kite, 1955). Following an open reduction, to treat hip dislocation, Rocha et al. (2010) obtained a reduced ROM in flexion (≥10˚) on four patients and increased abduction (≥10˚) in seven patients after a mean follow-up of 9.5 years. Bony surgery was performed to increase hip stability, reduce hip tightness and to correct hip deformity in 9 studies. After a reorientational proximal femoral osteotomy, H. J. van Bosse and Saldana (2017) found at a mean follow-up of 40 months that eighty-one hips that had a mean flexion contracture of 52˚ preoperatively improved by 35˚; 84 hips with a mean preoperative adduction of -20˚ improved by 42˚; 101 hips with a mean preoperative internal rotation of -16˚ improved by 35˚. In this same study, they also mentioned to perform soft-tissue surgery to treat mild hip contractures or to decrease flexion contractures allowing a less extreme osseous correction. Of the four studies including a lengthening procedure, two mentioned doing an adductor tenotomy to correct limitations of hip flexion or to reduce tension (H. J. van Bosse & Saldana, 2017; Wada et al., 2012), one did a tensor fasciae latae tenotomy for severe abduction contracture (Fassier et al., 2009) and one did not mention where the tenotomy has been done (Kroksmark et al., 2006). For muscle/tendon transfer, iliopsoas transfer was done on three patients (Hahn, 1985) and adductor transfer for progressive obliquity was done in one case (Herron et al., 1978); both studies did not provide a detailed description of the procedure used.

Casting was the most common conservative technique and was mostly used post-operatively for 5 weeks to 3 months (Aydin et al., 2016; Wada et al., 2012), and consisted of a hip Spica cast in most cases except for a Petrie cast used after osteotomy (H. J. van Bosse & Saldana, 2017). Binkiewicz-Glinska et al. (2013) treated a hip rotation deformity on a 3-week baby with plasters on the lower limbs combined with an



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aluminum crossbar and rehabilitation and found an improvement of 20˚ of hip rotation after 23 weeks of treatment. Rehabilitation was provided after surgery in three studies but no detail was provided and only Aydin et al. (2016) mentioned that the goal was to prevent hip stiffness. Rehabilitation was used as main treatment modality (Azbell & Dannemiller, 2015; Moore et al., 1990; Sala et al., 1996) or combined with casting (Binkiewicz-Glinska et al., 2013) among infants or a 9-year-old child, and led to ROM improvements or better hip position after a range of 11 weeks to 10 months of rehabilitation. After an intensive rehabilitation program (described earlier), Sala et al. (1996) reported passive gains from 30˚ of abduction to 15˚ of adduction and improved hip position from 45˚ external rotation to neutral. Pavlik harness was used in three studies (Akazawa et al., 1998; Ayadi et al., 2015; Wada et al., 2012) and Langeage was used by (Ayadi et al., 2015) with hip reduction in two of the four children treated with a Pavlik harness or by Langeage (Ayadi et al., 2015). Two studies reported avascular necrosis that may have been caused by the excessive force exerted by Pavlik harness wear (Akazawa et al., 1998; Wada et al., 2012). One study recommended using night splints for three months following hip surgery (Asif et al., 2004) and one study reported using an abduction splint (Yau et al., 2002). Orthoses were used in two studies, which consisted of a Seattle abduction orthosis on 20 hips after Spica cast immobilization (Szoke et al., 1996) or 10-month lively orthosis treatment (Moore et al., 1990) leading to a 10–15˚ increase of hip passive ROM. Asif et al. (2004) used successfully bracing to treat residual hip subluxation.

*Knee (n=27)*

Knee flexion contractures and knee extension contractures were mentioned in 20 and 19 studies respectively. In studies looking at both knee flexion and extension contractures, a higher rate of knee flexion contractures was found. Hall et al. (1983) reported that among 135 children with Amyoplasia, 87 had knee flexion contractures and 16 had knee extension contractures. Knee dislocations or subluxations were found in eight studies (Bharucha et al., 1972; Binkiewicz-Glinska et al., 2013; Borowski et al., 2008; Fucs, Svartman, de Assumpcao, & Lima Verde, 2005; Ghoreishi, BirjandiNejad, & Hallaj Moghadam, 2015; Kroksmark et al., 2006; Sud et al., 2013). Ayadi et al. (2015)



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reported an agenesis of the patella and displacement of the patella was reported in two studies (Binkiewicz-Glinska et al., 2013; Borowski et al., 2008). Ten studies used radiography to assess the position of the knee. Of these studies, DelBello and Watts (1996) measured the angle of the physis and the shaft of the femur on lateral radiograph and found a mean of 2° of flexion preoperatively on 12 patients with a mean age of 2.9 years and Fucs et al. (2005) noted five knees with recurvatum and six subluxated knees using Leveuf and Pais' radiographic classification. Ultrasound was used in two studies to assess the position of the patella in younger patients as unossified patella are not visible on radiographs (Binkiewicz-Glinska et al., 2013; Borowski et al., 2008) and to measure the size of the patella, the cartilaginous femoral sulcus angle (CSA) and the osseous femoral sulcus angle (OSA) of the femoral groove (Borowski et al., 2008). Patellar size of the affected side was 30% smaller when compared with the unaffected side. The average CSA was 161˚ (reference range, 146˚ ± 3.7˚) and the average OSA was 168˚ (references range 180 to 165˚) preoperatively. Borowski et al. (2008) also used MRI to assess the position of the patella. Knee flexion and extension contractures varied across studies from no ROM to full ROM (Bohm et al., 2013; Borowski et al., 2008; DelBello & Watts, 1996), as did the MMT ((Bohm et al., 2013; Moore et al., 1990). Using a hand-held dynamometer, Kroksmark et al. (2006) found muscle strength was reduced in flexion ($p=0.043$) and in extension ($p=0.001$) among non-ambulatory patients compared to house ambulators and community ambulators, and house ambulators had weaker knee flexion ($p=0.004$) and extension ($p<0.001$) compared to community ambulators. Knee extensor strength was strongly ($r=0.88$, $p<0.05$) and knee extension ROM was moderately ($r=0.59$, $p<0.05$) correlated to motor function.

To correct flexion or extension knee deformities, bony surgery was most common (e.g., supracondylar osteotomy, tibial osteotomy, distal femoral extension osteotomy). The Ilizarov technique was used to treat knee flexion contractures at a correction rate of 1-2 mm/day (Brunner, Hefti, & Tgetgel, 1997) or 0.65-1.38˚/ day (H. J. Van Bosse, Feldman, Anavian, & Sala, 2007) and provided significant ambulation gains (Yang, Dahan-Oliel, Montpetit, & Hamdy, 2010). Brunner et al. (1997) noted a higher recurrence rate of contractures for children under 10 years old, and Yang et al. (2010) reported that recurrences did not limit ambulatory gains. H. J. Van Bosse et al. (2007)



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mentioned that two patients had recurrences greater than 20˚. A knee fusion surgery was performed in one case after a recurrence (DelBello & Watts, 1996). Quadricepsplasty was performed mostly to correct extension contractures, and in some cases flexion contractures as well (Borowski et al., 2008; Ho & Karol, 2008). Fucs et al. (2005) and Ghoreishi et al. (2015) reported improved knee passive flexion from a pre-operative mean of 1.09˚ and 4.09˚ to a post-surgery mean of 67.30˚ and 70˚ with final quadriceps strength of grade 4 or 5 after a follow-up ranging between 2 years and 17.11 years after quadricepsplasty. Other procedures included hamstring and gastrocnemius lengthening, soft-tissue release and hamstring transfer with no specific results reported for them (Fassier et al., 2009; Hahn, 1985; Ho & Karol, 2008; H. J. Van Bosse et al., 2007; Yang et al., 2010).

Casting was used pre-operatively or as main treatment modality in seven studies. Fassier et al. (2009) found that early casting combined with PT was sufficient to correct deformities in 13 of 20 knees. Fucs et al. (2005) and Ghoreishi et al. (2015) reported that six months of manipulation and casting for hyperextended knees did not give successful results. Post-surgery casting lasting 3 to 10 weeks was mentioned in eight studies (Fucs et al., 2005; Ghoreishi et al., 2015). After surgery to correct extension contractures, casts were positioned between 30˚ and 90˚ of flexion (Borowski et al., 2008; Sud et al., 2013). Knee position was alternated between flexion and extension during every cast change (Fucs et al., 2005). Rehabilitation was administered pre-operatively or as main treatment to correct deformities or to prevent recurrence after surgery. Ghoreishi et al. (2015) provided one month of rehabilitation after quadricepsplasty with PT to increase ROM and strengthening followed by a home exercise program over three months. Binkiewicz-Glinska et al. (2013) used ultrasound to position the patella and the knee axis thus allowing for the progress of training, limiting, at the same time, the risk of the joint dislocation. Most of the time, rehabilitation consisted of stretching and manipulation, as well as strengthening (Azbell & Dannemiller, 2015; Ghoreishi et al., 2015). Orthoses were used to improve walking function, to prevent recurrence of contracture or as main treatment of knee flexion contractures. Gur, Erel, Yakut, Aksoy, and Uygur (2016) provided a serial orthotic treatment to decrease knee flexion contractures in two infants with ROM gains ranging 20˚ to 40˚. H. J. Van Bosse et al. (2007) and Gur et al. (2016)



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noted a recurrence of the contracture following discontinuing wearing orthoses. A 9.5-year-old girl was equipped for 10 months with a special resting orthosis containing gas springs to apply a constant extending moment. Her knee gained in extension ROM and lost in flexion, the arc of motion was considered more functional. Splinting was used after surgery, including three weeks of night splint with the knee in 30–45˚ of flexion after wire removal following an osteotomy (Sud et al., 2013) and to reduce contractures (Yang et al., 2010).

*Foot (n=31)*

Clubfoot (equinovarus) deformities were the most frequent deformity of the foot. Bartonek et al. (2011) noted 15–20˚ ankle contractures on eight of the sixteen participants. Brunner et al. (1997) reported preoperative equinus deformity ranged 10˚ to 60˚. The Dimeglio rating scale was used in four studies and clubfoot was initially graded between II and IV (Boehm, Limpaphayom, Alaee, Sinclair, & Dobbs, 2008). The Pirani grading scale was used by Iskandar, Bishay, Sharaf-El-Deen, and El-Sayed (2011) and they reported ten grade IV and five grade III deformities on fifteen feet. Of the 18 studies using radiography, detailed measures were reported in four studies (Choi, Yang, Chung, Cho, & Sohn, 2001; Iskandar et al., 2011; Segev, Ezra, Yaniv, Wientroub, & Hemo, 2008; Widmann, Do, & Burke, 2005). Initial standing antero-posterior angles were reported as follow: anterior talo 1st-metatarsal (0˚ to 76˚), talocalcaneal (0˚ to 33˚), calcaneo-5th metatarsal (15˚ to 48˚) and calcaneal-2nd metatarsal (20˚ to 60˚). Initial lateral angles were reported as follows: calcaneal-1st metatarsal (137˚ to 195˚), talocalcaneal (0˚ to 32˚), tibiocalcaneal (75˚ to 120˚) and talo-1st metatarsal (0˚ to 40˚). ROM measured at baseline was not mentioned in many studies but global ROM was between 15˚ to 30˚, dorsiflexion was about -15˚ to 30˚ and plantarflexion from 0˚ to 50˚ (Bartonek et al., 2007; Bohm et al., 2013; Choi et al., 2001). Using MMT, plantarflexor and dorsiflexor strength ranged from 0 to 5 (Bartonek et al., 2007; Bohm et al., 2013; Eriksson et al., 2015). Using a hand-held dynamometer, Kroksmark et al. (2006) reported that non-ambulators had weaker ankle dorsiflexors than community ambulators.

As for foot surgery, the Ilizarov procedure was used over 21 to 209 days (Brunner et al., 1997; Choi et al., 2001). Choi et al. (2001) reported after an average follow-up of

35 months that patients had good results but neglected use of brace and splint cause recurrence in one patient. Drummond and Cruess (1978) concluded that the most successful and reliable method of treating pes equinovarus was by arthrodesis. For soft tissue surgery, Ayadi et al. (2015) treated successfully six of the 11 clubfeet with postero-medial soft-tissue release and two of the three convex feet with soft-tissue release. Fassier et al. (2009) did a soft tissue release on 23 clubfeet to obtain plantigrade feet, six had lasting results. Widmann et al. (2005) did a posteromedial release in combination with serial cast and noted significant improvement in all lateral radiography more particularly in the tibiocalcaneal angle (131.18 to 82.25, p=0.00005) and talo-1st metatarsal angle (-26.45 to 0.75, p=0.00929) but not in antero-posterior radiography. According to muscle/tendon lengthening, almost all studies including this type of surgery did an Achilles tenotomy but few did a tenotomy of the calcaneal tendon or the tibialis posterior. Kroksmark et al. (2006) did lengthening of the Achilles tendon in nine patients and six who were operated early (mean = 1.5 months) had to undergo re-operation, when two of the three older children (> 1 years old) did not need another surgery. Other type of surgery included muscle/tendon transfer (e.g. tibialis anterior, tibialis posterior, peroneal brevis and hallucis longus) (Aroojis, King, Donohoe, Riddle, & Kumar, 2005; Drummond & Cruess, 1978; Mead et al., 1958; Schwering, 2005) and one study did a reduction of the talonavicular joint (Aroojis et al., 2005). Those surgeries were never the main procedure so any specific result was given for them.

In terms of conservative treatments, early casting (i.e., Ponseti technique) was commonly used to treat foot deformities. Cast start within the first year in almost all studies and was changed every 2 days to 2 weeks for a period of 6 weeks to 6 months (Aroojis et al., 2005; Hahn, 1985; Widmann et al., 2005). Ayadi et al. (2015) used Ponseti and found good results in four feet and recurrence in eight feet. Boehm et al. (2008) needed to do a tenotomy of the Achilles tendon to correct residual equinus deformity after serial casting management. They also mentioned that clubfeet in AMC required significantly more casting sessions than idiopathic clubfeet to achieve correction (6.9 ± 2.1 cast vs 4.5 ± 1.2 casts). Rehabilitation was successful to treat five valgus feet and three convex feet but 13 clubfeet required surgical correction following rehabilitation (Ayadi et al., 2015). Manipulation to stretch tight structures was done in combination with serial casting in some studies (Aroojis et al., 2005; Aydin et al., 2016; Hahn, 1985). Schwering (2005) promoted exercise to strengthen supinators and plantar flexors three times a day for 15 minutes by the parents. Sala et al. (1996) encouraged parents to provide rehabilitation at home including ankle dorsiflexion, eversion and abduction of the foot exercises but did not report any results. Orthoses were worn most of the time after surgery for a duration ranging from 1 year to full-time until skeletal maturity (Aroojis et al., 2005; Widmann et al., 2005). Splints or braces were worn to prevent recurrence of contractures after surgery for a period ranging from 4 years to 10 years when this detail was mentioned (Boehm et al., 2008; Hahn, 1985; Niki, Staheli, & Mosca, 1997). Two studies reported recurrence relating to non-compliance of the brace (Boehm et al., 2008; Choi et al., 2001). However, Boehm et al. (2008) reported that non-compliance may be caused by discomfort due to relapse or relapse may be caused by non-compliance.

## 4. DISCUSSION

Considering that the purpose of a scoping review is to expose gaps in current literature, this scoping review did not seek to evaluate treatment effectiveness. The aim of this scoping review was to identify how muscle and joint function are evaluated and treated among children with AMC. There were 87 studies included in this review. In general, surgery was well described but there was a lack of information about conservative treatment, especially rehabilitation despite rehabilitation playing a crucial role in preventing recurrences of contractures (Doi et al., 2011). As recommended by Hahn (1985), conservative treatments should be as fully documented as surgical procedures to communicate treatment approaches with clinicians and researchers, to evaluate treatment effectiveness by comparing the effect of similar treatments and to understand the impact of treatment on children with AMC and their families.

*Assessment*

Muscle strength was not assessed as often as joint position and ROM. In some studies, only initial or final results of muscle strength, ROM or joint position were reported. In order to evaluate treatment effectiveness, both baseline and follow-up outcomes are required. H. J. Van Bosse et al. (2007) reported their results following a



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mean follow-up of 52.7 months and mentioned useful information well as the age of surgery, type of AMC, length of follow-up, correction rate, ROM, arc of motion and the contracture's position. It is also important to clearly report the data, some studies reported negative knee ROM to describe flexion contractures or lack of knee extension, and other studies reported negative ROM in extension to describe hyperextension (Ghoreishi et al., 2015; Kroksmark et al., 2006). Studies should report whether ROM was measured actively or passively. Radiography was used to assess joint position and should be reported like it was done in Widmann et al. (2005) and Rocha et al. (2010), yet most studies did not provide specific radiographic data. Muscle strength should be measured especially when a muscle transfer or strengthening exercise program is done. It is preferable to use a dynamometer as it will allow evaluating improvement in muscle strength. It is important to understand that small improvements in ROM or muscle strength of children with AMC may lead to substantial functional improvements (Bernstein, 2002), and therefore should not be minimized. Validated tools and procedures for assessment of muscle and joint function should be used to detect minimally clinically important improvements.

*Management*

Providing evidence-based treatment to youth with AMC is essential. Many different types of surgical and conservative management were reported in this scoping review. Yet, important aspects needing reporting in order to establish best practice guidelines were lacking, such as age at treatment, type of AMC, details on the conservative treatment, and length of follow-up. Age at treatment is important to know for establishing the effectiveness of treatment because different viewpoints exist about the timing. Aydin et al. (2016) mentioned that performing surgery at a later age may be preferable because open reduction and femoral osteotomy procedures can be performed simultaneously to reduce the need for additional surgery. On the opposite hand, DelBello and Watts (1996) reported that although maintenance of correction post-operatively remains a problem, surgery should not be delayed until late childhood or adolescence as the benefits of walking, going to school, and interacting with peers far outweigh the risks of a potential second surgery later on. We agree with Hahn (1985) that mentioned the art



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of planning and providing a successful pediatric management program lies in applying the basic principles to the right child at the right times and many details should be known to make a decision about the right treatment. The diagnosis of multiple congenital contractures (i.e., Amyoplasia, or distal arthrogryposis) is important as some diagnoses may be more responsive to treatment than others (Aroojis et al., 2005). Also, conservative treatments are important to report as they have the potential to minimize recurrence of deformities (Gur et al., 2016; H. J. Van Bosse et al., 2007). However, it is not feasible to do a meta-analysis or a systematic review to calculate a pooled effect size or treatment effectiveness as most studies do not provide a detailed description of the treatment, and/or use a validated outcome measure. Initial condition (i.e. ROM, contractures) should be reported any time as treatment recommendations can differ (Palocaren et al., 2010). To our knowledge, only Miller and Sawatzky (2017) conducted a systematic review in AMC, which focused on upper limb surgery. They mentioned the lack of standardized assessments, the low level of evidence and the low rate of mid-term follow-up (> 2 years). Generally, surgical procedures were well described, and conservative treatments were not, except for casting. Conservative treatments are as important as surgery. Binkiewicz-Glinska et al. (2013) mentioned that daily exercises given by parents improve ROM and reduce the need for subsequent radical invasive corrections. Bernstein (2002) mentioned in his review that initial treatments of any contracture involve gentle stretching and range of motion exercises. However, the results found in this scoping review do not represent this statement as only 47% (33/70) reported providing conservative treatment before surgery. Only few studies mentioned providing muscle strengthening. We agree with Kroksmark et al. (2006) that reported that we should pay more attention to developing muscle strength in children with AMC having initially minimal strength. Hahn (1985) mentioned that non-operative treatments should be as fully documented as the operative notes. Rehabilitation, casts, braces, aids, and mobility devices need accurate reporting.

*Strengths and limitations*

This scoping review gives a better comprehension of the existing knowledge on the evaluation and treatment of muscle and joint function in children with AMC. All


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types of studies were included to give a better picture. On the other hand, studies were included regardless of methodological quality and therefore results should be interpreted with caution.

*Future research*

Future research should evaluate the effectiveness of therapies to guide the treatment plan for children with AMC. To achieve this, a systematic review and meta-analysis should be used; however, this type of knowledge synthesis requires a homogenous population, procedure and outcome measure for pooling purposes, which is often not the case in these studies. There is a need for longer follow-up periods with validated tools and a detailed description of the interventions provided to share knowledge and standardize treatment. Follow-ups should be until completion of the growth as contractures can recur when the child is growing up. Clinicians and researchers should therefore create guidelines to standardize procedures, timing, and choice of outcome measures used to evaluate the effectiveness of treatment. Further studies should assess the effectiveness of some promising interventions like the use of exoskeleton or nerve transfer as they seemed to give good results but it was on few children only. As AMC does not affect only the joint, attention should also be paid to the evaluation of muscle function and to improve it by strengthening. Only a few papers included in this scoping review have received financial support. Funding for studies in this area could promote higher levels of evidence leading to evidence-based practice for children with AMC.

## 5. Conclusion

In conclusion, this scoping review exposes gaps in the current literature on AMC and guides directions for future research. Muscles and joints are assessed and treated in multiple ways in youth having AMC but further studies are needed to establish what is best for them. These findings provide guidance as to how muscle and joint function are evaluated and treated among children with AMC, and should be used to inform the development of consensus-based guidelines for best practice in this population. Studies should provide the diagnosis leading to multiple congenital contractures, use validated

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measures, describe the conservative interventions in greater details, and include follow-up periods extending to skeletal maturity to advance research and inform evidence-based treatment of children with AMC.

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## 7. DECLARATION OF INTEREST

The authors have no conflicts of interest to declare.

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For Peer Review

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**TABLES**

Table 1: List of selection criteria

| | Inclusion criteria | Exclusion criteria |
|---|---|---|
| **Population: age** | 0 to 21 years | > 21 years |
| **Population: diagnosis** | Any type of AMC | Does not provide separate results for AMC in studies with more than one condition |
| **Study design** | Qualitative, quantitative or mixed methods | No exclusion based on study design |
| **Assessment** | Use of tools or/and outcome measure | Muscle and joint function not measured |
| **Treatment** | Described surgical procedure or rehabilitation treatment addressing 5 domains | Described rehabilitation intervention or surgical procedure addressing treatment to another domain |
| **Language** | English and French | English or French translation were not included |

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Table 2: Number of studies (n) according to different assessment types for every joint

| **Location** | **Range of motion** | **Position** | **Muscle strength** | **Muscle and bone composition** | **Total** |
|---|---|---|---|---|---|
| Head/Jaw | 5 | 4 | 2 | 1 | 12 |
| Shoulder | 7 | 8 | 2 | 0 | 17 |
| Elbow | 27 | 16 | 14 | 0 | 57 |
| Hand | 16 | 23 | 5 | 0 | 44 |
| Spine | 0 | 8 | 0 | 0 | 8 |
| Hip | 19 | 24 | 6 | 0 | 49 |
| Knee | 20 | 26 | 11 | 0 | 57 |
| Foot | 12 | 30 | 4 | 0 | 46 |
| General | 0 | 1 | 1 | 2 | 4 |
| Total | 106 | 140 | 45 | 3 | 294 |

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Table 3: Tools and methods to assess muscle and joint function

| **Joint/bones** | **n** | **Muscle** | **n** | **Nerve** | **n** | **Motion Analysis** | **n** |
|---|---|---|---|---|---|---|---|
| Radiography | 41 | MMT | 18 | CNS microhistopathology | 1 | Vicon camera system | 4 |
| Goniometry | 15 | Biopsy | 2 | Nerve biopsy | 1 | Force plate | 1 |
| Tomography | 2 | Histology | 3 | Electrical stimulation | 1 | Real time motion analysis system | 1 |
| CT scan | 2 | Creatine kinase | 2 | | | | |
| DEXA | 1 | EMG | 8 | | | | |
| MRI | 1 | Electron microscopy | 2 | | | | |
| | | Weight | 2 | | | | |
| | | Dynamometer | 3 | | | | |

Legend:

CNS microhistopathology: Central nervous system microhistopathology

DXA: Dual-energy x-ray absorptiometry

EMG: Electromyography

MMT: Manual muscle testing

MRI: Magnetic resonance imaging

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Table 4: List of grading scales to assess muscles or joints

| Name | Designed to | Number of studies |
|---|---|---|
| Ankle-hindfoot clinical rating system | Assess foot function | 1 |
| Bleck classification | Assess the position of the forefoot | 1 |
| Coleman classification | Assess the severity of vertical talus deformity | 1 |
| Dimeglio grading system | Assess severity of the foot deformity | 4 |
| Gilbert's grading system | Assess thumb function | 1 |
| Graf's classification | Assess developmental dysplasia of the hip | 1 |
| Gruel classification | Assess motion at the hip | 1 |
| Kalamchi & MacEwen classification | Assess avascular necrosis at the hip | 2 |
| Leveuf Pais classification | Assess the articular position of the knee | 1 |
| McKay's score | Clinical assessment of the hip | 1 |
| Medical Council Research Grade | Assess manually muscle power on a scale of 0 to 5 | 6 |
| Modified clubfoot outcome grading system | Assess functional results, appearance, and patient satisfaction of the ankle and foot | 2 |
| Oxford scale | Assess manually muscle strength on a scale of 0 to 5 | 1 |
| Pirani Score | Assess the severity of clubfoot deformity | 1 |
| Severin | Assess the radiographic results of the hip dislocation | 4 |
| St Clair hip score | Assess hip function | 1 |
| Tönnis grading system | Assess developmental dysplasia of the hip with radiography | 2 |
| Walker score | Assess developmental flatfoot | 1 |

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Table 5: Number of studies (n) according to different surgery types for every joint

| | **Soft Tissue (n=55)** | **Bony Surgery (n=57)** | **Muscle/tendon lengthening (n=35)** | **Muscle/nerve transfer (n=37)** | **Reduction (n=17)** |
|---|---|---|---|---|---|
| **Head/ Jaw (n=4)** | • Release of the fibrous adhesions (n=1) | • Condylectomy<br>• Coronoidectomy<br>• Temporo-mandibular arthroplasty (n=2) | No studies | No studies | • Temporal eminence reduction (n=1) |
| **Shoulder (n=2)** | No studies | • Humeral osteotomy<br>• Derotation osteotomy (n=2) | No studies | No studies | No studies |
| **Elbow (n=41)** | • Capsulotomy<br>• Posterior elbow joint release (n=13) | • Shoulder fusion<br>• Humeral rotational osteotomy (n=4) | • Tricepsplasty<br>• V-Y lengthening of the distal triceps (n=7) | • Latissimus dorsi flexorplasty<br>• Pectoralis major transfer<br>• Tricep transfer<br>• Deltoid transfer<br>• Pedicled latissimus dorsi bipolar muscle transfer<br>• Steindler flexorplasty<br>• Clark transfer<br>• Gracilis muscle transfer | No studies |

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| | | | | | |
|---|---|---|---|---|---|
| | | | | • Fascicular ulnar nerve transfer (n=17) | |
| **Hand (n=37)** | • Z-plasty<br>• Web space deepening<br>• Volar wrist release<br>• Release of the palmar fascia<br>• Capsulotomy<br>• Skin Flap (n=12) | • Chondrodesis<br>• Carpal resection<br>• Carpectomy<br>• Ulnar osteotomy<br>• Navicular excision<br>• Dorsal wedge osteotomy<br>• Thumb fusion<br>• Metacarpophalangeal fusion<br>• Wrist fusion (n=10) | • Lengthening of the flexor tendon<br>• Lengthening of the flexor pollicis longus tendon<br>• Lengthening of the flexor pollicis profondus tendon (n=4) | • Opponensplasty<br>• Transfer of the extensor carpi<br>• Transfer of the flexor carpi ulnaris<br>• Transfer of a sublimis tendon<br>• Transfer of the wrist flexor (n=11) | No studies |
| **Spine (n=3)** | No studies | • Harington instrumentation<br>• Spinal fusion<br>• Osteotomy<br>• Anterior and posterior bony fusion (n=3) | No studies | No studies | No studies |
| **Hip (n=36)** | • Soft tissue release (n=6) | • Acetabuloplasty<br>• Girdlestone procedure<br>• Proximal femoral osteotomy<br>• Salter innominate | • Hamstring lengthening<br>• Quadriceplasty<br>• Tensor fasciae latae tenotomy (n=4) | • Iliopsoas transfer<br>• Adductor transfer (n=2) | • Open reduction<br>• Closed reduction (n=15) |

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| | | | | | |
|---|---|---|---|---|---|
| | | osteotomy<br>• Shortening osteotomy<br>(n=9) | | | |
| **Knee (n=32)** | • Posterior soft tissue release<br>• Soft tissue release<br>(n=9) | • Femoral shortening<br>• Supracondylar osteotomy<br>• Tibial osteotomy<br>• Extending osteotomy<br>• Anterior distal femoral stapling<br>• Ilizarov procedure<br>• Knee fusion<br>(n=12) | • Quadricepsplasty<br>• Hamstring lengthening<br>• Gastrocnemius lengthening<br>(n=10) | • Hamstring transfer<br>(n=1) | No studies |
| **Foot (n=46)** | • Tarsal tunnel release<br>• Postero-medial soft-tissue release<br>• Posterior release<br>• Plantar fasciotomy<br>• Soft-tissue release<br>(n=14) | • Triple Arthrodesis<br>• Talectomy<br>• Tarsal resection<br>• Decancellation of the cuboid, calcaneus and talus<br>• Toes fusion<br>• Wedge osteotomy<br>• Ilizarov procedure<br>• Osteotomy<br>(n=15) | • Tenotomy of the Achilles tendon<br>• Tenotomy of the calcaneal tendon<br>• Tenotomy of the tibialis posterior<br>• Tibialis anterior lengthening<br>• Lengthening of Achille tendon<br>(n=12) | • Transfer of the tibialis posterior<br>• Transfer of the tibialis posterior<br>• Transfer of the peroneal brevis<br>• Transfer of the hallucis longus<br>• Talectomy<br>(n=4) | • Reduction of the talonavicular joint<br>(n=1) |

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Table 6: Number of studies (n) according to different conservative interventions for each joint

| **Location** | **Splint** | **Brace** | **Cast** | **Orthosis** | **Harness** | **Rehabilitation** | **Traction** | **Exoskeleton** | **Immobilisation** | **Total** |
|---|---|---|---|---|---|---|---|---|---|---|
| Head/jaw (n=3) | 0 | 1 | 0 | 0 | 0 | 2 | 0 | 0 | 0 | 3 |
| Shoulder | 0 | 0 | 0 | 0 | 0 | 4 | 0 | 1 | 0 | 5 |
| Elbow | 7 | 1 | 7 | 2 | 0 | 15 | 0 | 1 | 1 | 34 |
| Hand | 12 | 0 | 8 | 1 | 0 | 11 | 0 | 0 | 1 | 33 |
| General upper limb | 2 | 0 | 1 | 1 | 0 | 3 | 0 | 1 | 0 | 8 |
| Spine | 0 | 4 | 2 | 1 | 1 | 2 | 2 | 0 | 0 | 10 |
| Hip | 2 | 1 | 11 | 2 | 3 | 7 | 0 | 0 | 0 | 26 |
| Knee | 4 | 0 | 12 | 10 | 0 | 11 | 0 | 0 | 0 | 37 |
| Foot | 7 | 4 | 21 | 9 | 0 | 13 | 0 | 0 | 0 | 54 |
| General lower limb | 0 | 1 | 0 | 1 | 0 | 1 | 0 | 0 | 0 | 3 |
| General | 2 | 0 | 0 | 1 | 0 | 5 | 0 | 0 | 1 | 9 |
| **Total** | 36 | 12 | 62 | 28 | 4 | 74 | 2 | 3 | 3 | 224 |

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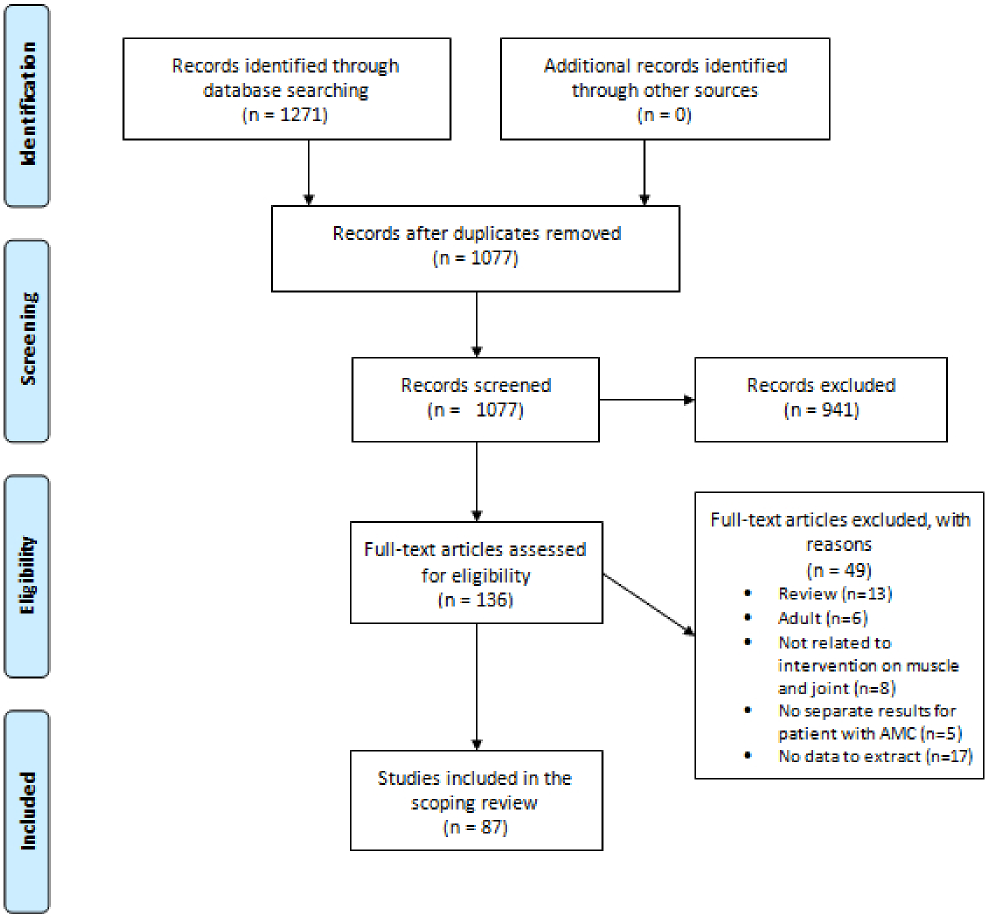


Figure 1: PRISMA flow chart

201x191mm (72 x 72 DPI)

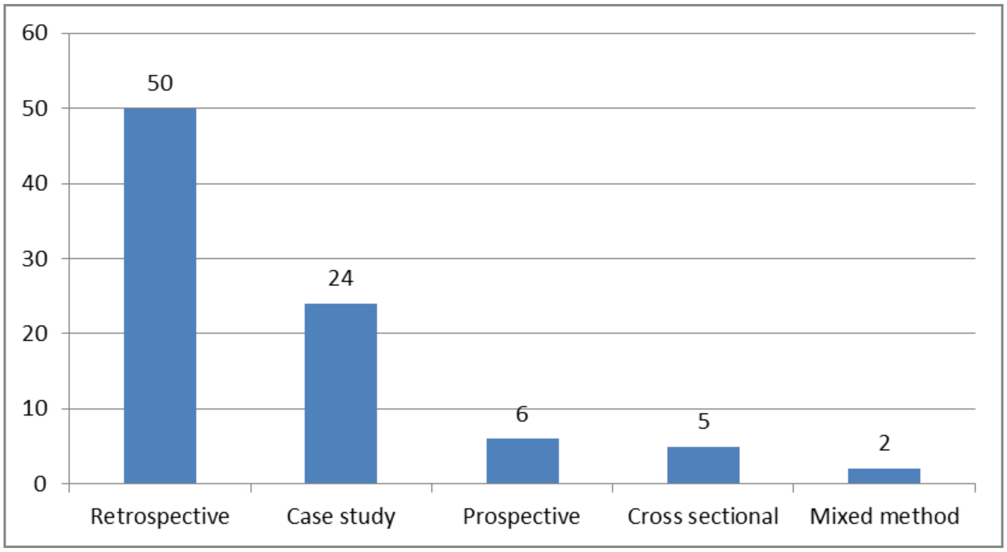


Figure 2: Study design of included studies in the scoping review (n=87)