Assessments and interventions for youths with arthrogryposis multiplex congenita

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August 2020

A thesis submitted to McGill University in partial fulfillment of the requirements of the degree of Master of Science in Experimental Surgery

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List of abbreviations

AFO	Ankle-foot orthosis
APPT	Adolescent and Pediatric Pain Tool
AMC	Arthrogryposis multiplex congenita
CNESST	Commission des normes, de l’équité, de la santé et de la sécurité du travail
CNS	Central nervous system
CSA	Cartilaginous femoral sulcus angle
DXA	Dual-energy x-ray absorptiometry
EMG	Electromyography
FO	Foot orthosis
GAS	Goal Attainment Scale
HEP	Home exercise program
ICC	Intraclass correlation coefficient
KAFO	knee-ankle-foot orthosis
MMT	Manual muscle testing
MRI	Magnetic resonance imaging
MSK	Musculoskeletal
OSA	Osseous femoral sulcus angle
OT	Occupational therapist
PAQ-A	Physical Activity Questionnaire for Adolescents
PODCI	Pediatrics Outcomes Data Collection Instrument
PRISMA-ScR	Preferred reporting items for systematic reviews and meta-analyses extension for scoping reviews
PRT	Physical Rehabilitation Therapist
PT	Physical therapist
ROM	Range of motion
SAAQ	Société de l’assurance automobile du Québec
SHC	Shriners Hospital for Children

Abstract

Background: Arthrogryposis multiplex congenita (AMC) is a term used to describe the presence of congenital joint contractures in two or more body areas, and affects 1 in 4300 live births. AMC is associated with limited joint range of motion (ROM) and muscle weakness leading to functional limitations in daily activities. Interventions for AMC aim at improving function and mobility. Different types of assessments and interventions are used with this population and different viewpoints exist about the best timing and type of intervention.

Methods: The first phase of the thesis consisted of a scoping review aiming at identifying how muscle and joint functions are evaluated and treated among children with AMC. A search in four different databases was conducted. Identified articles were reviewed according to the following selection criteria: (a) published in French or English; (b) include children (0 to 21 years); (c) include any type of AMC; (d) describe muscle and joint function; (e) describe surgical procedure or conservative intervention. The general information, methods, results, and conclusions were extracted and findings were synthesized.

The second phase of the thesis consisted in developing a 12-week individualized home exercise program (HEP) for youths with AMC and delivered remotely. At baseline and at the end of the intervention, participants completed online standardized questionnaires related to physical activity (PAQ-A), functional status (PODCI) and pain (APPT). A physical therapist and an occupational therapist performed a remote assessment with participants. ROM were measured with a virtual goniometer. The Goal Attainment Scale (GAS) was used to identify individualized goals to develop a 12-week HEP. Different feasibility criteria such as withdrawal rates, compliance to the HEP and to the follow-ups were collected. Pre- and post-results on the PAQ-A, PODCI and APPT and the results from the GAS were used to explore the effectiveness of the intervention.

Results: In the scoping review, 87 studies were included. Joints were studied in all studies whereas muscles were studied in 30 studies. Bony surgery was the most common surgery and rehabilitation the most common conservative intervention. It was found that surgery was well detailed when conservative interventions were not and that better studies were needed to develop guidelines.

In the telerehabilitation study, seven participants (median: 16.9 years) completed the intervention. Results showed that, besides poorer than expected recruitment rates, telerehabilitation is a feasible approach to deliver therapeutic intervention in this population. For the effectiveness, based on the GAS, 12 of the 15 objectives set were achieved. Significant improvements were observed in the pain and comfort domain of the PODCI (p=0.08) as well as in the PAQ-A (p=0.03).

Conclusions: Performing research in individuals with rare disorders is difficult, among other reasons, because of the remoteness of the patients. The use of telerehabilitation is a step forward to improve access to adjunct therapies otherwise not possible to develop and deliver. Telerehabilitation will also help developing innovative research projects with sufficient power and sample size through facilitating multicenter studies which will increase representation of findings and lead to developing guidelines for key stakeholders. Exercise has the potential to help individuals with AMC achieve their goals and should be examined in a randomized control trial.

Abrégé

Introduction : L'arthrogrypose multiple congénitale (AMC) est un terme utilisé pour décrire la présence de contractures articulaires dans au moins deux régions du corps. L'AMC affecte 1 naissance sur 4300. L'AMC est caractérisé par une limitation dans l'amplitude articulaire et une faiblesse musculaire entraînant des limitations fonctionnelles dans les activités quotidiennes. Les interventions pour l'AMC visent à améliorer la fonction et la mobilité. Différents types d'évaluations et d'interventions sont utilisés avec cette population et différents points de vue existent à propos du meilleur moment et types d'intervention.

Méthodes : La première phase du projet consistait en un examen de la portée visant à identifier comment les fonctions musculaires et articulaires sont évaluées et traitées chez les enfants avec l'AMC. Une recherche dans quatre bases de données différentes a été effectuée. Les articles identifiés ont été examinés selon les critères de sélection suivants : (a) publiés en français ou en anglais ; (b) inclut les enfants (de 0 à 21 ans) ; (c) inclut tout type d'AMC ; (d) décris la fonction musculaire et articulaire ; (e) décris une intervention chirurgicale ou conservatrice. Les informations générales, méthodes, résultats et conclusions ont été extraits et synthétisés.

La deuxième phase du projet consistait à développer un programme d'exercices à domicile individualisé de 12 semaines pour les jeunes avec l'AMC et dispensé à distance. Au début et à la fin de l'intervention, les participants ont rempli en ligne des questionnaires standardisés en lien avec l'activité physique (PAQ-A), la fonction (PODCI) et la douleur (APPT). Un physiothérapeute et un ergothérapeute ont effectué une évaluation à distance avec les participants. Les amplitudes articulaires ont été mesurées avec un goniomètre virtuel. L'échelle de réalisation des objectifs (GAS) a été utilisée pour identifier des objectifs individualisés pour développer le programme d'exercice. Différents critères de faisabilité tels que les taux d'abandon, le taux de participation au programme et aux suivis ont été recueillis. Les résultats pré- et post-intervention pour le PAQ-A, le PODCI et l'APPT et les résultats du GAS ont été utilisés pour explorer l'efficacité de l'intervention.

Résultats : Dans l'examen de la portée, 87 études ont été incluses. Les articulations ont été étudiées dans toutes les études, tandis que les muscles ont été étudiés dans 30 études. La chirurgie osseuse était la chirurgie la plus courante et la réadaptation était l'intervention conservatrice la plus fréquente. Il a été constaté que les chirurgies étaient bien détaillées alors que les interventions

conservatrices ne l’étaient pas. Dans l’étude sur la téléréadaptation, sept participants (médiane : 16,9 ans) ont terminé l’intervention. Les résultats ont montré qu’excepté les taux de recrutement plus faibles que prévu, la téléréadaptation est une approche faisable pour fournir une intervention thérapeutique à cette population. Pour l’efficacité, 12 des 15 objectifs fixés avec le GAS ont été atteints. Des améliorations significatives ont été observées dans le domaine de la douleur du PODCI ($p = 0.08$) ainsi que dans le PAQ-A ($p = 0.03$).

Conclusions : La recherche avec des patients atteints de maladies rares est difficile, entre autres, en raison de l’éloignement des patients. L’utilisation de la téléréadaptation est un pas vers l’avant pour améliorer le développement des thérapies d’appoint (ex : programme d’exercice) autrement difficile à offrir. Cette approche aidera également à développer des projets de recherche innovants avec une puissance et une taille d’échantillon suffisantes ce qui augmentera la représentation des résultats et conduira à l’élaboration de lignes directrices pour les principales parties prenantes. L’exercice a le potentiel d’aider les personnes avec l’AMC à atteindre leurs objectifs et devrait être évalué dans un essai randomisé contrôlé.

Acknowledgements

I would like to thank the two most important people who helped me complete this thesis: my supervisor, Dr. Louis-Nicolas Veilleux, and my co-supervisor, Dr. Noémi Dahan-Oliel. Thank you for taking the time to have discussions, to support my work and to review and answer my questions even when I was last minute or you were very busy. Thank you for all the opportunities you gave me and everything you did for me to make sure that I could reach my full potential and to succeed in my Master studies.

Louis-Nicolas, you are an extraordinary, caring and outstanding person. You believed in me from the beginning until the end. I will forever be grateful that you convinced me to study at McGill University even when I lacked the confidence to study in my second language. It was not easy studying in my second language and it made you more revision to perform, but I am very thankful that you have embarked me on this project that changed my life. Louis-Nicolas, thanks to you, I am very proud of all the progress I have made since I met you in 2017. I am grateful for all your support whether it was academically, financially or mentally. I learned much more than research skills, as you also taught me about the importance of balancing graduate studies, work and personal life.

Noémi, I am so glad that you accepted to include me in your project. You are an inspiring person. Thank you for your listening and always being present when I needed you. You helped through my emotional rollercoasters as I like to call. I am grateful for all you did especially when it got challenging. You helped me bring this thesis further with all your ideas. You always knew how to push me further and I am very grateful of that as I accomplished so many things with you.

I also owe a special thanks to everyone in the research team who contributed to the success of this project. Thank you to Kevin Caporuscio, Gabriela Marino Merlo, Rita Yap, Jessica Collins, Caroline Elfassy, Dr. Jacquelyn Marsh, Dr. Bonita Sawatzky and Dr. Hamdy. Thank you to Dr. Emmanouil Rampakakis who advised me on the statistical component of this project. Thank you to Mariam Morgan for taking this project further with the infographic and podcast. Thank you to Guylaine Bedard to have made many beautiful posters and images.

I am also thankful to all the youths and their families for their participation in this project. You gave meaning to my research and you were my main source of motivation throughout this project.

I am grateful to the Canadian Musculoskeletal rehab research network that provided funding support for the telerehabilitation project. I would also like to acknowledge the Défi Pierre Lavoie Foundation which awarded me a master's scholarship (2019–2020).

Thank you to all my Shriners colleagues, who became like a family in the past two years and contribute to my success. Special thanks to Enrique, Jean-Pascal, Clement and Louis-Nicolas, you are all so funny and made me laugh on bad days. Thank you to Carla and Souad who were always present to support me and listen to all my adventures.

To all my friends, more particularly Anne-Marie and Mariane, I will always remain thankful for your moral support and all your encouragements when I needed them. You were always present to celebrate all my little accomplishments, such as my first poster presentation, my first publication or even when I just move a little forward in my project during the covid-19 pandemic. Special thanks to Don who was always there to answer all my questions, who helped me improve my English and succeed in class (even in stats), who listened to me complain about everything and who encouraged me to take breaks and relax.

Last but not least, I would like to thank my family for their support and their confidence in me. Your encouragement helped me persevere in this project and helped me believe I can achieve anything I want.

Contribution of Authors

This thesis consists of two published manuscripts and one manuscript that has been submitted to the International Journal of Environmental Research and Public Health for the special issue "Evaluating Telerehabilitation for Patients across the Lifespan."

Chapter 1: Marianne wrote the chapter and Dr. Louis-Nicolas Veilleux and Dr. Noémi Dahan-Oliel reviewed the chapter.

Chapter 2: Marianne did the data collection, data analysis and interpretation and wrote the manuscript. Kevin Caporuscio collected part of the data and reviewed the manuscript. Dr. Dahan-Oliel supervised the students and participated in the data collection, analysis and interpretation and revised the manuscript. Dr. Hamdy and Dr. Veilleux supervised the students and took part in the data interpretation and manuscript revision.

Chapter 3: Dr. Dahan-Oliel initiated, planned, and received funding for this study and wrote the study protocol. Marianne Gagnon wrote the paper. Jessica Collins, Caroline Elfassy, Gabriela Marino Merlo, Dr. Marsh, Dr. Sawatzky, Rita Yap, Dr. Hamdy, Dr. Veilleux and Dr. Dahan-Oliel reviewed the manuscript.

Chapter 4: Dr. Dahan-Oliel initiated and received funding for this study and wrote the study protocol. Dr. Dahan-Oliel, Dr. Veilleux, Dr. Sawatzky and Dr. Marsh develop the methodology. Marianne Gagnon, Jessica Collins, Caroline Elfassy, Gabriela Marino Merlo and Rita Yap were involved in the data collection. Marianne Gagnon, Dr. Veilleux and Dr. Dahan-Oliel participated in data analysis and data interpretation. Marianne Gagnon wrote the manuscript. Dr. Veilleux, Jessica Collins, Caroline Elfassy, Gabriela Marino Merlo, Dr. Marsh, Dr. Sawatzky, Rita Yap, Dr. Hamdy and Dr. Dahan-Oliel reviewed the manuscript. Dr. Rampakakis gave statistical advised and reviewed the manuscript.

Chapter 5: Marianne wrote the chapter. Dr. Veilleux and Dr. Dahan-Oliel reviewed the chapter.

Chapter 6: Marianne wrote the chapter. Dr. Veilleux and Dr. Dahan-Oliel reviewed the chapter.

Chapter 1: Introduction

1. Rationale

Shriners Hospital for Children-Canada (SHC) is a world leader in specialized care for children with rare congenital musculoskeletal disorders. A rare disorder is characterized by its low prevalence in the population and defined as "a disorder that affects less than 1 in 2000 people in a population" (1, 2). Approximately 7,000 rare diseases and disorders have been reported around the world and nearly 75% of these disorders affect children (3). Musculoskeletal conditions include conditions that affect the locomotor system (i.e. muscles, bones, joints and associated tissues such as tendons and ligaments). Musculoskeletal conditions are typically characterized by pain and limitations in mobility, dexterity and functional ability (4). Given the complexity of these disorders, patients with rare musculoskeletal disorders often need subspecialized health care that may not be readily available in their local community. In 2019, 4.73% of patients actively followed at the SHC-Canada came from other Canadian provinces and 1.99% came from other countries. Even patients living in Quebec province may need to travel long distances to come at the SHC-Canada (e.g. Gaspésie). Living far from the subspecialized health care center entails direct and indirect costs to the patient and their family. Families need to take days-off from work and school (indirect costs) and have travel costs to the subspecialized health care center (direct costs) (5, 6). From a clinical perspective, children living far from subspecialized health care centers and/or in remote areas may not have similar access to regular therapeutic interventions (7). This means that new intervention approaches and technologies are needed to increase access to sub-specialized care for patients living remotely from the hospital. Telerehabilitation, which consists of remote delivery of rehabilitation services using telecommunication technologies (8), may offer a potential solution to overcome the distance challenge. The benefits and challenges related to a telerehabilitation approach will be detailed in the next section.

The feasibility of telerehabilitation has been reported in different populations and for different purposes (9-12). A multicenter randomized clinical trial across Quebec compared face to face rehabilitation (n=100) with the same intervention program provided by telerehabilitation (n=98) for patients who undergone a total knee arthroplasty. Looking at pain, stiffness and function, a 2% difference in those scores between telerehabilitation and face-to-face rehabilitation was reported suggesting similar care efficiency (8). When used with home-exercise program, telerehabilitation brings several advantages. For example, it is known that home exercise program (HEP) without supervision shows participation rates to the training program varying between 11% and 37% depending on how many times patients should perform exercises in a week (13). When a HEP was combined with telerehabilitation, a participation rate of 76% was found (14). Another study showed that cost is reduced by using telerehabilitation compared to home visits when patients live over a distance of 30 km from the health care center (15). Telerehabilitation may serve as a helpful strategy for continuing monitoring and management of a patient allowing face-to-face intervention, including relatives (e.g. spouse) in a familiar environment and as part of everyday life (16). Telerehabilitation has taken a step forward and has started to be used to make remote evaluations. Remote evaluations for range of motion (ROM), muscle testing and mobility assessment have been shown to have high inter- and intra-rater reliability when conducted remotely with healthy adults (10). As an example, knee flexion and extension can be measured via a virtual goniometer (17). In addition to these benefits, patients and clinicians have good satisfaction with this approach. A study providing remote occupational therapy hand assessment reported that 94% of the patients were comfortable being on videoconference, 100% agree that telerehabilitation will allow easy access to healthcare and 89% agree that telerehabilitation will save them time and money. On the clinician side, 94% were satisfied with the level of service the telerehabilitation system allowed them to provide, 83% found they were able to satisfactorily and competently assess and treat the client (18).

On the other hand, some aspects can limit the usability of telerehabilitation. In certain cases, patients recently experienced a modification of their physical or cognitive status. It may become hard to manipulate telerehabilitation technology (e.g. the keyboard) when they are not adapted to their new condition (19). In another study, limited technological skills, missing face-to face contact with medical staff and other patients, no “hand on” real time contact and reimbursement were reported as limitations (16). Despite those limitations, telerehabilitation has the potential to improve health care access and reduce the specific costs associated within the context of rare disorders.

One of the most frequently represented rare musculoskeletal disorders managed at SHC-Canada is arthrogryposis multiplex congenita (AMC). AMC is an umbrella term that represents a group of conditions characterized by joint contractures present in at least two different body areas at birth (20). The overarching goal of my thesis was to pilot a telerehabilitation approach to deliver a regular therapeutic intervention for young individuals with AMC to achieve individualized goals.

2. Objectives

The aim of this prospective pilot project is to provide an assessment and an exercise intervention at home for youths with AMC using telerehabilitation. Specifically, the objectives are to 1) evaluate the feasibility of using telerehabilitation for delivery of a home-exercise program (HEP) in youths with AMC; 2) explore the potential effectiveness of the HEP on joint mobility, muscle strength and pain. Relying on patients' satisfaction, recruitment and compliance, we hypothesize that telerehabilitation will be a feasible approach to provide an exercise intervention as an adjunct mode of service delivery. As part of the feasibility objective, the inter-rater reliability of using a virtual goniometer to assess ROM will also be assessed. We expect to observe a compliance rate for our exercise intervention ranging between the compliance rate reported for an unsupervised HEP (11-37%) (13) and a supervised HEP (76%) (14). As our HEP is semi-supervised, we hypothesize a compliance rate of 50%. For the second objective, we hypothesize improvements in muscle strength, joint mobility and pain. However, we expect to observe improvements at different extents for each participant in-line with their individualized goals.

Chapter 2: Literature Review

Connecting text

In May 2016, at the SHC-Canada, Dr. Reggie Hamdy and Dr. Noémi Dahan-Oliel established the only Canadian multidisciplinary AMC clinic for children between birth and 21 years of age. In December 2016, Dr. Dahan-Oliel organized a knowledge exchange day at the SHC-Canada funded by a Canadian Institute of Health Research SPOR grant and more than 120 different key stakeholders (youth, adults with AMC, parents and clinicians) participated and shared their priorities regarding research in AMC. Among the different priorities that arose during this exchange day, one of the highest priorities was to have more information on potential interventions aiming at improving physical function. Another important priority was related to exercise that can lead to improvement. This research project seeks to address these stakeholders' priorities. The first step in addressing those research priorities is to establish the current state of knowledge on assessments and surgical and conservative interventions and on the effect of exercise on patient's functional status. A scoping review was therefore completed to reach that first step. This scoping review is presented in this chapter.

Manuscript 1: Muscle and Joint Function in Children Living with Arthrogryposis Multiplex Congenita: A Scoping Review

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Gagnon M, Caporuscio K, Veilleux L-N, Hamdy R, Dahan-Oliel N. Muscle and joint function in children living with arthrogryposis multiplex congenita: A scoping review. *Am J Med Genet Part C*. 2019; 1–17. https://doi.org/10.1002/ajmg.c.31726

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 1,271 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.

KEYWORDS

arthrogryposis multiplex congenita, joint, management, muscle, pediatric

1. Introduction

Arthrogryposis affects from 1 in 3000 to 1 in 4300 live births (Lowry, Sibbald, Bedard, & Hall, 2010). Arthrogryposis multiplex congenital (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 muscle weakness. These factors contribute to the formation of contractures (Ferguson & Wainwright, 2013; Hall, 2014). Current clinical treatment to overcome these contractures are based on a conservative approach (i.e., nonsurgical) as well as surgical management, both aiming at improving function, and mobility (Oishi et al., 2017; Van Bosse et al., 2017).

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 with AMC reported that their ability to participate in meaningful activities and roles are affected by their environment, such as the home, school, and community. 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 are 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.

1. 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's (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).

1.1 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.

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 five domains	Described rehabilitation intervention or surgical procedure addressing treatment to another domain
Language	English and French	English or French translation were not included

1.2 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 10 research articles 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.

2. Results

2.1 Search strategy and study selection

The search strategy yielded a total of 1,271 articles. Following removal of duplicates, 1,077 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.

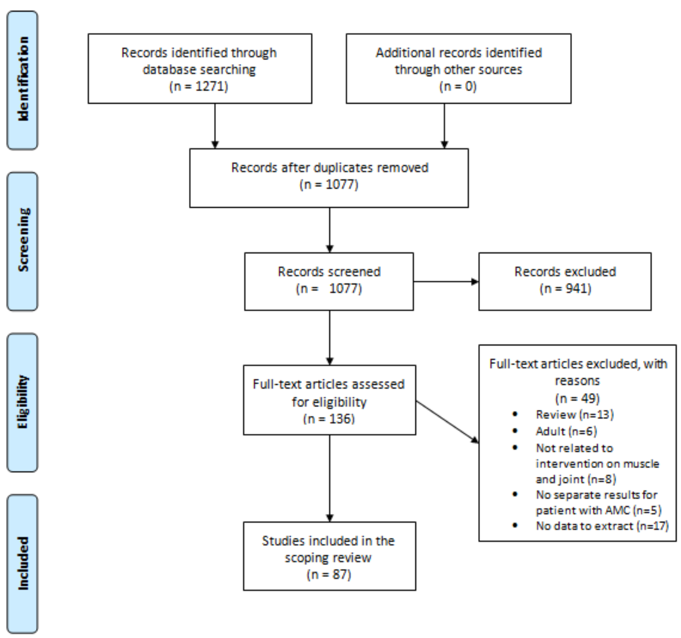

Figure 1: PRISMA flow chart

2.2 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 articles 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.

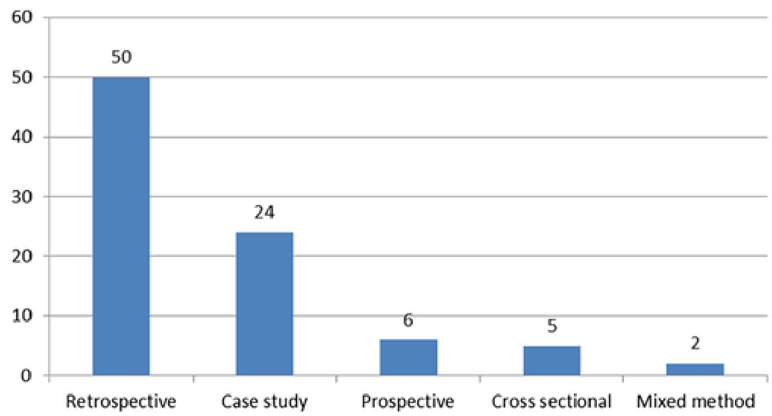

Figure 2: Study design of included studies in the scoping review (n=87)

2.3 Sample characteristics

The 87 included studies result in a total of 1,426 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 10 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.

2.4 Assessment and management of joint and muscle function

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. Bone mineral density was measured in one study (Spencer et al, 2010), with higher bone density being correlated with increasing ambulatory function. ROM was measured actively (n = 14), passively (n = 12), or both (n = 19). Description of the arc of motion was present in 23 studies. Muscle strength was most commonly assessed using the 5-point medical research council grade 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.

Table 2. Number of studies according to different assessment types for every joint location

Location	Range of motion	Position	Muscle strength	Muscle and bone composition
Head/Jaw	5	4	2	1
Shoulder	7	8	2	0
Elbow	27	16	14	0
Hand	16	23	5	0
Spine	0	8	0	0
Hip	19	24	6	0
Knee	20	26	11	0
Foot	12	30	4	0
General	0	1	1	2

Table 3. Number of studies (n) using different 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				

Abbreviations: CNS microhistopathology, central nervous system microhistopathology; DXA, dual-energy x-ray absorptiometry; EMG, electromyography; MMT, manual muscle testing; MRI, magnetic resonance imaging.

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 the 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	Assess the hip clinically	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

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 9 of 13 patients, including abnormal predominance of type I fibers in five children over 10 months of age, and type II fibers in three 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 et al., 2011; Bartonek, Eriksson, & Gutierrez-Farewik, 2007; Bohm, Dussa, Multerer, & Doderlein, 2013; Eriksson, Bartonek, Ponten, & Gutierrez- Farewik, 2015). Eriksson et al. (2015) found that all AMC groups (i.e., wearing knee-ankle-foot orthoses (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 the importance of an adequate orthotic support. Bohm et al. (2013) found that AMC patients' gait showed excessive range in thorax obliquity in 10 of the 18 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). Applying heel heights adequate to each individual's orthopedic 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 (PT and OT), 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 articles 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.

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 • Coronoidectomy • Temporo-mandibular arthroplasty (n=2)	No studies	No studies	• Temporal eminence reduction (n=1)
Shoulder (n=2)	No studies	• Humeral osteotomy • Derotation osteotomy (n=2)	No studies	No studies	No studies
Elbow (n=41)	• Capsulotomy • Posterior elbow joint release (n=13)	• Shoulder fusion • Humeral rotational osteotomy (n=4)	• Tricepsplasty • V-Y lengthening of the distal triceps (n=7)	• Latissimus dorsi flexorplasty • Pectoralis major transfer • Tricep transfer • Deltoid transfer • Pedicled latissimus dorsi bipolar muscle transfer • Steindler flexorplasty • Clark transfer • Gracilis muscle transfer • Fascicular ulnar nerve transfer (n=17)	No studies
Hand (n=37)	• Z-plasty • Web space deepening • Volar wrist release • Release of the palmar fascia • Capsulotomy • Skin Flap (n=12)	• Chondrodesis • Carpal resection • Carpectomy • Ulnar osteotomy • Navicular excision • Dorsal wedge osteotomy • Thumb fusion • Metacarpophalangeal fusion • Wrist fusion (n=10)	• Lengthening of the flexor tendon • Lengthening of the flexor pollicis longus tendon • Lengthening of the flexor pollicis profondus tendon (n=4)	• Opponensplasty • Transfer of the extensor carpi • Transfer of the flexor carpi ulnaris • Transfer of a sublimis tendon • Transfer of the wrist flexor (n=11)	No studies
Spine (n=3)	No studies	• Harington instrumentation • Spinal fusion • Osteotomy • Anterior and posterior bony fusion (n=3)	No studies	No studies	No studies
Hip (n=36)	• Soft tissue release	• Acetabuloplasty • Girdlestone procedure	• Hamstring lengthening	• Iliopsoas transfer	• Open reduction

	(n=6)	• Proximal femoral osteotomy • Salter innominate osteotomy • Shortening osteotomy (n=9)	• Quadriceplasty • Tensor fasciae latae tenotomy (n=4)	• Adductor transfer (n=2)	• Closed reduction (n=15)
Knee (n=32)	• Posterior soft tissue release • Soft tissue release (n=9)	• Femoral shortening • Supracondylar osteotomy • Tibial osteotomy • Extending osteotomy • Anterior distal femoral stapling • Ilizarov procedure • Knee fusion (n=12)	• Quadricepsplasty • Hamstring lengthening • Gastrocnemius lengthening (n=10)	• Hamstring transfer (n=1)	No studies
Foot (n=46)	• Tarsal tunnel release • Postero-medial soft-tissue release • Posterior release • Plantar fasciotomy • Soft-tissue release (n=14)	• Triple Arthrodesis • Talectomy • Tarsal resection • Decancellation of the cuboid, calcaneus and talus • Toes fusion • Wedge osteotomy • Ilizarov procedure • Osteotomy (n=15)	• Tenotomy of the Achilles tendon • Tenotomy of the calcaneal tendon • Tenotomy of the tibialis posterior • Tibialis anterior lengthening • Lengthening of Achille tendon (n=12)	• Transfer of the tibialis posterior • Transfer of the tibialis posterior • Transfer of the peroneal brevis • Transfer of the hallucis longus • Talectomy (n=4)	• Reduction of the talonavicular joint (n=1)

Table 6. Number of studies according to different conservative interventions for each joint

Location	Splint	Brace	Cast	Orthosis	Harness	Rehabilitation	Traction	Exoskeleton	Immobilization
Head/jaw	0	1	0	0	0	2	0	0	0
Shoulder	0	0	0	0	0	4	0	1	0
Elbow	7	1	7	2	0	15	0	1	1
Hand	12	0	8	1	0	11	0	0	1
General upper limb	2	0	1	1	0	3	0	1	0
Spine	0	4	2	1	1	2	2	0	0
Hip	2	1	11	2	3	7	0	0	0
Knee	4	0	12	10	0	11	0	0	0
Foot	7	4	21	9	0	13	0	0	0
General lower limb	0	1	0	1	0	1	0	0	0
General	2	0	0	1	0	5	0	0	1

2.4.1 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 90^0, L 70^0) and lateral cervical flexion (R 40^0, L10^0) 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). 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 35^0 of improvement of the left lateral cervical flexion (10 to 45) after a 9-month PT and OT intervention. Epstein and Wittenberg (1987) reported several surgical procedures including an intraoral 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 followup. 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).

2.4.2 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, & Dastur (1972). Studies using ROM measured flexion (0–120°), extension (0–20°), abduction (40–180°), internal rotation (excessive to 90°), and external rotation (0–70°) (Babik et al., 2016; Kroksmark et al., 2006; Sala, Rosenthal, & Grant, 1996). Shoulder muscle 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.

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.

2.4.3 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 articles assessing muscle strength before elbow surgery reported no bicep strength except two who found a bicep strength of 2–3/5 (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 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; Doyle et al, 1980; Van Heest et al, 1998). After a combination of tricepsplasty and tricep transfer and an average follow-up of almost 5 years among 23 elbows, muscle strength ranged from 0 to 5 on a scale of five (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 10 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 6 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, Van Heest et al. (2008) reported an improvement in the arc of passive motion of all 29 elbows from an average of 32° (range, 0–75°) preoperatively to an average of 66° (range, 10–125°) postoperatively. Wall et al (2017) describes distal humerus external rotation osteotomy to improve grasp pattern among nine children with Amyoplasia with positive results.

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 was indicated to reduce recurrences of contractures. Casts were kept for a period ranging from 3 to 6 weeks in a flexion position (Atkins et al., 1985; Gagnon, Fogelson, & Seyfer, 2000). Splints were used for a period ranging from 4 weeks to 6 months (Goldfarb et al., 2004; Mennen, 1993). Post-surgery rehabilitation included muscle re-education and passive and active mobilization. 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) 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).

2.4.4 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, 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; Van Heest & Rodriguez, 2013). Post-surgery, cast were used most of the time until bone consolidation (4 to 9 weeks) (Burgess & Robbe, 2012; 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 Amyoplasia 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 Amyoplasia group. With regard to rehabilitation, Sala et al. (1996) provided an early intensive rehabilitative and splinting treatment from 4 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 16 hr/day. An improvement of wrist extension (40–45) and active flexion of her right wrist and digits was reported at the final assessment.

2.4.5 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 with a vital capacity of 0.33 liters (L), indicative of severe restrictive ventilator impairment in a 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 3 years after surgery. Moreover, atelectasis of the right lobe was resolved, vital capacity improved to 0.53 L and posture was improved leading to a stable gait. Herron et al. (1978) did 10 hip surgeries 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 postoperative 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 preoperative curve (75 and 108°) underwent an osteotomy combined with a spinal fusion and had better postoperative 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 2 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.

2.4.6 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 assessing hip position with specific measures, such as the acetabular index (28–50 before surgery) and the center-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 IV (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 comparing muscle strength in children with different ambulation levels.

Non ambulators and house ambulators had significantly weaker hip flexors than community ambulators, hip extensors and hip abductors.

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 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 center-edge angle from −41° preoperatively to 18° at a mean follow-up of 4 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 nine studies. After a reorientational proximal femoral osteotomy, Van Bosse and Saldana (2017) found at a mean follow-up of 40 months that 81 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 performed 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 (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 postoperatively 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 (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 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 harness was used by Ayadi et al. (2015) with hip reduction in two of the four children treated with a Pavlik or Langeage harness (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 3 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 orthosis treatment (Moore et al., 1990) leading to a 10–15° increase of hip passive ROM. Asif et al. (2004) used bracing successfully to treat residual hip subluxation.

2.4.7 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) 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 reduced muscle strength in knee flexion and extension among non-ambulatory patients compared to house ambulators and community ambulators. House ambulators had weaker knee flexion and extension compared to community ambulators. Knee extensor strength was strongly and knee extension ROM was moderately 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 (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. Van Bosse et al. (2007) 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 preoperative 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; Van Bosse et al., 2007; Yang et al., 2010).

Casting was used preoperatively 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 6 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 preoperatively or as main treatment to correct deformities or to prevent recurrence after surgery. Ghoreishi et al. (2015) provided 1 month of rehabilitation after quadricepsplasty with PT to increase ROM and strengthening followed by a home exercise program over 3 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. Van Bosse et al. (2007) and Gur et al. (2016) 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 3 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).

2.4.8 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 16 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 10 grade IV and five grade III deformities on 15 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 anteroposterior angles were reported as follows: anterior talo firstmetatarsal (0–76°), talocalcaneal (0–33°), calcaneo-fifth metatarsal (15–48°) and calcaneal-second metatarsal (20–60°). Initial lateral angles were reported as follows: calcaneal-first metatarsal (137–195°), talocalcaneal (0–32°), tibiocalcaneal (75–120°) and talofirst metatarsal (0–40°). ROM measured at baseline was not mentioned in many studies but global ROM was between 15 and 30°, dorsiflexion was about −15–30° 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 at an average follow-up of 35 months that patients had good results, but neglected use of brace and splint caused recurrence in one patient. Drummond and Cruess (1978) concluded that the most successful and reliable method of treating pes equinovarus was by arthrodesis. Other bony surgeries included, as example, talectomy (Green et al., 1984; Hsu et al., 1984) and navicular excision (Mubarak & Dimeglio, 2011). 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) and talo-first metatarsal angle (−26.45 to 0.75) but not in anteroposterior 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 types 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 not the main procedure described in those respective articles, therefore specific results were not described.

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 4 feet and recurrence in 8 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 noncompliance of the brace (Boehm et al., 2008; Choi et al., 2001).

However, Boehm et al. (2008) reported that noncompliance may be caused by discomfort due to relapse or relapse may be caused by noncompliance.

3. 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, 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.

3.1 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. Van Bosse et al. (2007) reported their results following a mean follow-up of 52.7 months and mentioned useful information well as the age at 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.

3.2 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 the 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 for evaluating treatment effectiveness 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 of planning and providing a successful pediatric management program relies 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 appropriate treatment. The diagnosis of multiple congenital contractures (i.e., Amyoplasia, distal arthrogryposis or other) 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; 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 did not provide a detailed description of the treatment, and/or did not use a validated outcome measure. Initial condition (i.e., ROM, contractures) should be reported 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 welldescribed, 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.

3.3 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 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.

3.4 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 exoskeletons or nerve transfers. As AMC does not affect only the joint, attention should also be paid to the evaluation of muscle function. Only a few articles 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.

4. 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 youth with AMC. 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 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.

Acknowledgment

Much appreciation is given to Koorosh Semsar-Kazerooni and Alexa Cirillo for screening titles, abstracts, and full texts, and to Dr. Mohammed AlOtaibi for his advice on surgical procedures.

Conflict of interest

The authors have no conflicts of interest to declare.

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Studies included in the scoping review (n=87) are indicated with asterisk.

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Chapter 3: Methodology

Connecting text

The findings from the scoping review (chapter 3) demonstrated that little is known about the effect of exercise on muscles and joints in youths with AMC. Keeping in mind that exercise was a priority needing to be addressed as shared by individuals with AMC during the exchange day in 2016, we decided to pilot the delivery of a home exercise program. However, as AMC is rare, we had to identify an innovative method of delivery to provide regular follow-ups to youths with AMC living across Canada. Therefore, we decided to use a telerehabilitation approach and pilot the feasibility of this approach. These findings can then guide the development of a larger study to assess the effectiveness of a home-based exercise program in youth with AMC. The protocol for this project is presented in this chapter.

Manuscript 2: A Telerehabilitation Intervention for Youths with Arthrogryposis Multiplex Congenita: Protocol for a Pilot Study

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Gagnon M, Collins J, Elfassy C, Marino Merlo G, Marsh J, Sawatzky B, Yap R, Hamdy R, Veilleux LN, Dahan-Oliel N. A Telerehabilitation Intervention for Youths with Arthrogryposis Multiplex Congenita: Protocol for a Pilot Study. *JMIR Res Protoc*. 2020; 9 (6): e18688. doi: 10.2196/18688. URL: http://www.researchprotocols.org/2020/6/e18688/

Abstract

Background: Arthrogryposis multiplex congenita (AMC) is characterized by joint contractures present in at least two body areas. In addition to these contractures, individuals with AMC can have decreased muscle mass, leading to limitations in activities of daily living. Exercise has the potential to maintain or improve the range of motion and muscle strength. However, this type of intervention necessitates frequent follow-ups that are currently difficult to provide for youths with AMC because they often live far from a specialized hospital. To overcome this distance challenge, telecommunication technologies can be used to deliver rehabilitation remotely, which is called telerehabilitation. The study protocol for one such type of rehabilitation will be presented in this paper.

Objective: This pilot study aims to (1) evaluate the feasibility of using telerehabilitation to provide a home exercise program for youths with AMC, and (2) assess the effectiveness of a home exercise program.

Methods: A total of 10 youths aged 8-21 years with AMC will be recruited. The intervention consists of a 12-week individualized home-based exercise program delivered remotely using telerehabilitation. At baseline, youths will complete the Physical Activity Questionnaire for Adolescents and the Pediatrics Outcomes Data Collection Instrument to assess pain, function, and level of physical activity. During the first telerehabilitation meeting, the rehabilitation therapists will measure range of motion using a virtual goniometer and assess the youth's functional level. The therapists will then use the Goal Attainment Scale to set objectives and develop the individualized intervention. Follow-ups will occur every 3 weeks to make sure exercises are performed safely and to progress the exercises when needed. At the end of the 12-week intervention, rehabilitation therapists will re-evaluate the youth using the same outcome measures as the initial evaluation. The youths will be asked to complete the same questionnaires, with the addition of questions about their satisfaction regarding the intervention. Nonparametric and descriptive statistics will be used to evaluate the feasibility and effectiveness.

Results: Ethics approval was obtained in October 2018. Recruitment and data collection started in January 2019 and was completed in May 2020.

Conclusions: This pilot study will help us learn how a large-scale project may work in practice to improve outcomes in physical activity, pain, and function, and goal attainment among youths with AMC, thus informing a future clinical trial.

KEYWORDS: telerehabilitation; arthrogryposis multiplex congenita; physical therapy; occupational therapy

Introduction

Arthrogryposis multiplex congenita (AMC) or arthrogryposis is a term representing a heterogeneous group of over 400 disorders characterized by congenital contractures present in at least two different body joints [1]. AMC is rare and affects 1 in 3000 to 1 in 4300 live births [2]. Amyoplasia and distal arthrogryposis are the most common types and combined together, they represent about 50%-65% of all AMC diagnoses [3,4]. Contractures, which are defined as the limitation of passive movement of a specific joint, can affect the joints of the upper and/or lower limbs, as well as the spine and jaw [1]. Contractures can be caused by an increase of connective tissue around the joints, joint fibrosis replacement of muscle, muscle atrophy, or articular deformities [1]. AMC is typically nonprogressive, as no new contractures appear during an individual's life. However, contractures can recur after intervention or worsen over time [5]. In addition to these contractures, individuals with AMC may present with decreased muscle mass and bone density, bone deformities, and pain, leading to activity limitations and participation restrictions in activities of daily living such as feeding, dressing, mobility, and sports [3,6-9]. Early interventions such as stretching, splinting, bracing, casting, rehabilitation, and surgical procedures have been shown to augment and maintain range of motion and strength, and therefore, to promote independence in daily activities [10].

Rehabilitation can improve physical function and maintain gains after surgery [5]. Structural changes of the joint surface in AMC further limit range of motion when the joint is not used [10]. Besides, a positive association has been reported between knee and hip muscle strength and motor function [11], suggesting that rehabilitation exercises can maintain or increase range of motion and preserve muscle strength for optimal motor function. Currently, most interventions in AMC, specifically rehabilitation, occur in early childhood and their frequency decreases during school-age and adolescent years, despite new challenges arising during these transition periods [5,12].

Rehabilitation for school-aged children focuses mostly on body functions and structure, which does not always correspond to the youth's specific needs, such as participating in activities [12].

As AMC is rare, youths are mainly treated in subspecialized health care centers, which may be geographically distant from where they live. Therefore, clinicians face an important challenge in implementing regular exercise interventions. Novel intervention approaches and technologies are needed to increase access to subspecialized care for youths with AMC across geographical boundaries. Telerehabilitation, defined as "an innovative way to deliver rehabilitation services remotely using information and telecommunication technologies" [13], can be used to overcome this challenge. Some aspects can limit the usability of telerehabilitation, such as the lack of direct contact with participants, difficulties with manipulation of technology for individuals with physical limitations, or having a poor internet connection [14,15]. Nevertheless, telerehabilitation has been studied with different clinical populations (eg, total knee arthroplasty or vascular surgery patients) and was found to be as effective as face-to-face interventions [13], save travel time [16], and reduce cost for people living at a distance of 30 km or more from the health care center [17]. In the pediatric population, telerehabilitation has been used to provide various interventions (physical therapy, psychology, and speech language therapy) for different populations (patients with acquired brain injury, autism spectrum disorder, or cerebral palsy) [18]. Despite the potential benefits of telerehabilitation, there is a lack of research on its use for youths with physical impairments [18]. Therefore, the purpose of this study is to pilot the delivery of a home-based exercise program (HEP) for youths with AMC using telerehabilitation. Specifically, this pilot study aims to (1) evaluate the feasibility of using telerehabilitation to provide a physical assessment and to deliver an HEP for youths with AMC; and (2) explore the potential effectiveness of the HEP on goal attainment, physical activity, pain, and function. The provision of an HEP through telerehabilitation will provide an unprecedented opportunity for service equity to this vulnerable population, regardless of geographical location. The methodology of this study is reported in this paper.

Methods

Study Design

This is a pilot study to evaluate the feasibility and effectiveness of a telerehabilitation intervention for youths with AMC.

Participants Eligibility

Youths aged between 8 and 21 years with a confirmed clinical diagnosis of multiple congenital contractures, or AMC, will be invited to participate. Inclusion criteria include the ability to communicate in English or French and residence in Canada.Youths living in another country will be excluded as rehabilitation therapists participating in this study hold only a Canadian professional license. To align with postintervention precautions, those having undergone a recent surgery (ie, 3 months for soft tissue and 6 months for bony surgery) will be excluded. Youths with cognitive deficits or unstable health will also be excluded, to ensure participants can take part in the HEP. To assess the level of cognition, medical records will be reviewed for the presence of central nervous involvement or intellectual impairment. In addition, the treating physician, therapists, and parents will be consulted to determine eligibility based on type of schooling and ability to follow instructions. No restriction will be made about the degree of severity of contractures or physical impairment.

Recruitment

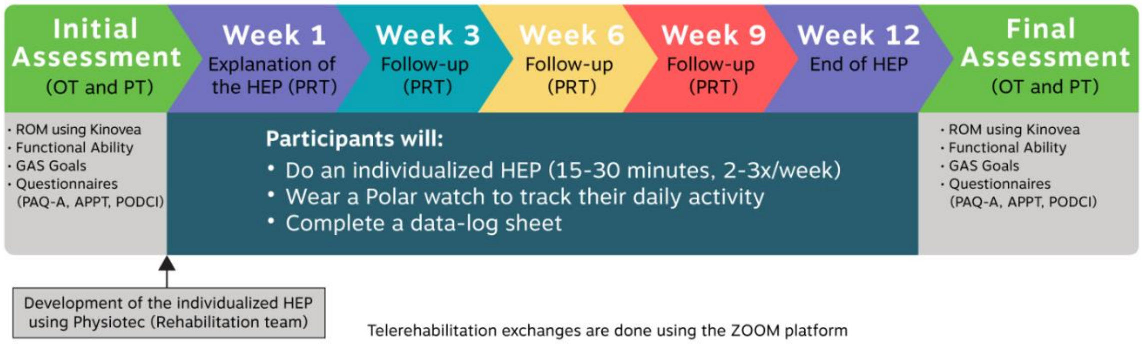
Youths with AMC followed at Shriners Hospital for Children - Canada (SHC-C) will be recruited during their clinic visit. Potential youths with the appropriate clinical diagnosis who do not have an upcoming visit to SHC-C will be contacted by postal mail and phone. For youths with AMC who are not followed at SHC-C, an advertisement describing the study will be posted on social media of a Canadian AMC support group (ie, Facebook). Those interested in participating will be asked to contact the clinical research coordinator for more information. Prior to participating in the study, informed consent will be sought from parents as well as from youths aged 14 years and older, as per provincial regulations. Youths between the ages of 8 and 13 years will be asked to provide assent. For these younger participants, the research team will encourage parents to be available during the telerehabilitation sessions to provide support during the sessions and throughout the HEP. As we expect to complete this study on 10 youths, 13 youths will be recruited to account for a 30% dropout rate [19].

Intervention

A rehabilitation team including a physical therapist, an occupational therapist, and a physical rehabilitation therapist will collaborate to provide the intervention. A physical rehabilitation therapist is defined as a professional among the interdisciplinary team whose role is to obtain necessary prerequisite information from a leading physical therapist in order to develop treatment plans and provide appropriate interventions to reduce activity limitations and participation restrictions.

The intervention will consist of a 12-week HEP. Prior to the assessment with the therapists, youths will complete an online questionnaire consisting of the Physical Activity Questionnaire for Adolescents (PAQ-A), the Pediatrics Outcomes Data Collection Instrument (PODCI), and demographic questions. The occupational therapist and physical therapist will conduct the initial assessment of each youth using ZOOM Pro (Zoom Video Communications, Inc), a videoconferencing platform allowing an encrypted connection. During this assessment, the therapists will perform an active joint range-of-motion (AROM) assessment of the upper and lower limbs, evaluate overall function, assess pain using the Adolescent and Pediatric Pain Tool (APPT), and establish individualized goals with each youth using the Goal Attainment Scale (GAS). The information gathered through this initial assessment will be used by the rehabilitation team to develop an individualized 12-week HEP. A week after the initial assessment, the physical rehabilitation therapist will exchange with the youth using ZOOM to explain and deliver the HEP as well as ensure comprehension and safe execution of the exercises. Youths will be asked to perform their HEP three times a week for approximately 15-30 minutes each time. They will be sent a physical activity monitor (POLAR watch A370) by postal mail and will be instructed to wear it on their wrist during HEP sessions, as it will be used to capture heart rate and the duration of the exercise sessions. Youths will have the possibility to wear the physical activity monitor at all times if they desire. If needed for the HEP, exercise materials such as resistant elastic bands or TheraPutty will also be sent to them by postal mail. A follow up with the physical rehabilitation therapist will be provided through ZOOM every 3 weeks (ie, at weeks 3, 6, and 9) to address any questions and to adjust the HEP as needed. At the end of the 12-week intervention, the occupational therapist and physical therapist will re-evaluate the youth using the same outcome measures as the initial evaluation. Youths will be asked to complete the same questionnaire, with the addition of questions about their satisfaction regarding the intervention. Figure 1 provides a summary of the intervention. The outcome measures used in this study are described in the following sections. In addition, parents will be asked to complete a cost questionnaire (direct and indirect costs incurred in relation to their child’s condition), which will be described elsewhere as it extends beyond the scope of this study.

Figure 1: A summary of the 12-week telerehabilitation intervention.



APPT: Adolescent and Pediatric Pain Tool; GAS: Goal Attainment Scale; HEP: home-based exercise program; OT: occupational therapist; PAQ-A: Physical Activity Questionnaire for Adolescents; PODCI: Pediatrics Outcomes Data Collection Instrument; PRT: physical rehabilitation therapist; PT: physical therapist.

Measurement

Feasibility

Operationalization

The source of recruitment (eg, in clinic, postal mail, phone, or social media), recruitment and withdrawal rates, compliance to the HEP and to the telerehabilitation meetings, and missing data will be calculated to determine feasibility of the intervention for youths with AMC. The compliance to the HEP will be measured using the data from the activity monitor as well as from a data-log sheet on which youths will have to record when they perform their exercise program. Refer to Table 1 for the operationalization of the feasibility criteria.

Table 1. Operationalization of the criteria to evaluate feasibility.

Parameters	Definition	Criteria/examples
Source of recruitment	The method used to recruit each youth.	In clinic, postal mail, phone, social media
Recruitment rates	From a list of patients with AMC[a] followed, the number of eligible youths will be determined. Those that are reachable will be accounted.	≥ 50% of eligible and reachable youths
Withdrawal rates (Before the intervention)	Youths who will consent to participate but will withdraw before the start of the intervention will be accounted.	≤ 20% of the youths who consent
Withdrawal rates (During the course of the intervention)	Youths who will complete at least one telerehabilitation meeting and will decide to withdraw afterwards will be counted. The time points when they decide to withdraw will be collected as well as the reason, if applicable.	≤ 30% of the youths who start the intervention
Completion rates	The number of youths who will complete all 6 telerehabilitation meetings out of those who have consented.	≥ 50% of the youths who consent
Compliance to the HEP[b]	The amount of time youths performed their HEP will be collected using a participant-completed log sheet and data from the physical activity monitor.	≥ 50% of compliance to the HEP
Compliance to the telerehabilitation meetings	The number of meetings cancelled the same day among the meetings that occurred, as well as lateness to the meetings. Lateness is defined as joining the meeting 15 minutes or more after the scheduled time.	≤ 15% of the meetings
Missing data	The number of questionnaires not completed and the number of unusable range of motion data.	≤ 10% for each outcome
Technical issues	Problems that will arise and disrupt or delay the meeting or possibly prevent the telerehabilitation meeting from taking place.	Echo voices, connection, image quality

[a]AMC: arthrogryposis multiplex congenita.

[b]HEP: home-based exercise program.

Cost

The total amount of time spent during the telerehabilitation intervention by the physical therapist, occupational therapist, and the physical rehabilitation therapist providing the assessment and the HEP as well as their time outside the direct intervention with the youth (planning the HEP and writing the report) will be recorded. The cost of the ZOOM plan and of the federal professional licenses for the occupational therapist and physical therapist will be reported.

Satisfaction

Satisfaction regarding the telerehabilitation intervention, the assessment with the therapists, and the HEP will be evaluated in the final online questionnaire with open-ended questions and using a 5-point Likert scale.

Effectiveness

GAS

The GAS will be administered by the occupational therapist with the youth during the initial online assessment to set individualized goals with the youths and to calculate the extent to which their goals are met at the end of the 12-week HEP [20].

AROM

AROM will be measured to provide an overview of the youths' ability to build the HEP. Screenshots of each youth performing specific movements during the initial and final assessment will be taken and then AROM will be measured using a virtual goniometer (Kinovea). This method has been shown to be feasible on 10 healthy adult participants [21]. AROM of the following joints will be measured as degrees of movement: shoulder (abduction, flexion, and extension), elbow (flexion and extension), forearm (pronation and supination), wrist (flexion and extension), hip (flexion, extension, internal rotation, and external rotation), knee (flexion and extension), and ankle (dorsiflexion and plantar flexion). Some movements are expected to be difficult to measure with a virtual goniometer because of the wrong plane of movement, so we will report the following AROM as full, limited, or absent: finger and thumb (flexion, extension, abduction, and adduction), shoulder (internal and external rotation), and hip (abduction). When possible, AROM will be measured in positions that allow being with and without the effect of gravity to estimate muscle strength. Although passive range of motion is usually taken clinically, it will not be measured in this context of telerehabilitation.

PODCI

This questionnaire was developed by the American Academy of Orthopaedic Surgeons and the Pediatric Orthopaedic Society of North America to measure functions in the following dimensions among children aged 2-18 years with musculoskeletal conditions: upper extremity functioning, transfers and basic mobility, sports and physical function, and comfort/pain [9]. The four dimensions are computed together to give a global functioning score, and in addition, a separate scale evaluating happiness with physical condition is provided. The PODCI response scales use a 3- to 6-point format. The internal consistency is high and the test–retest reliability is excellent for all subscale scores [22]. The PODCI was shown to be sensitive to change in function over time in 74 children with amyoplasia [9].

PAQ-A

This questionnaire measures the general levels of physical activity in the last 7 days. The PAQ-A is a self-administered questionnaire developed for typically developing high-school-age youths. This questionnaire has shown a moderately high concurrent validity when compared with an activity monitor and a good internal consistency [23]. The PAQ-A provides a summary physical activity score derived from 8 items, each scored on a 5-point scale. The final mean of the 8 items is reported to classify the level of physical activity [24].

Adolescent Pediatric Pain Tool

This tool assesses current pain for children and adolescents between ages 8 and 17 years with different conditions such as cancer and orthopedic and traumatic injuries. The APPT shows good validity, test–retest reliability, and sensitivity. The questionnaire uses a visual analog scale, a list of 42 qualitative words describing pain, and 2 body diagrams on which the child has to circle the location of the pain [25]. A summary of the outcomes to assess the effectiveness is presented in Table 2.

Table 2. Summary of outcomes used to determine effectiveness.

Tools / questionnaire	What does it assess?
Adolescent Pediatric Pain Tool (APPT)	• Pain
Goal Attainment Scale (GAS)	• Progress of the individualized goals
Physical Activity Questionnaire for Adolescents (PAQ-A)	• Level of physical activity
Pediatrics Outcomes Data Collection Instrument (PODCI)	• Upper extremity function • Transfers and basic mobility • Sports and physical function • Comfort and pain • Global function • Happiness
Range of motion (ROM)	• Shoulder: abduction, adduction, flexion and extension • Elbow: flexion and extension • Forearm: pronation and supination • Wrist: flexion and extension • Hip: flexion, extension, internal rotation and external rotation • Knee: flexion and extension • Ankle: dorsiflexion and plantar flexion

Statistical Analyses

Given this is a pilot study with a small sample size and a heterogeneous population, nonparametric and descriptive statistics will be used. To assess feasibility, the compliance to the HEP and to the telerehabilitation meetings, recruitment and completion rates, missing data, and closed-ended questions on satisfaction will be analyzed using descriptive statistics and compared with the established feasibility criteria. Table 1 describes the feasibility criteria for operationalization that will be used in the study. Youths' experience about use of technology and overall satisfaction with the program will be reported as collected in the open-ended questions. The cost of the telerehabilitation intervention (ie, therapists' time, federal professional license, and teleconferencing cost), recruitment source, and technical issues experienced during the program will be described. For the effectiveness of the HEP, raw GAS scores will be converted to GAS T-scores for each youth and for the global HEP. Preintervention and postintervention data of the range of motion, APPT, PAQ-A, and PODCI will be compared using the Wilcoxon signed-rank test to measure change among the same youth.

Results

Administrative site approval was obtained from the Department of Medical Research at Shriners Hospital for Children (CAN1806) in July 2018. Ethics approval was provided by the McGill University Faculty of Medicine Institutional Review Board (#A08-B38-18B) in October 2018. Recruitment and data collection started in January 2019 and was completed in May 2020.

Discussion

Overview

The proposed telerehabilitation pilot study is designed to provide an HEP to youths with AMC. Establishing the feasibility of this study has the potential to inform service delivery models using technology, thereby overcoming geographical barriers, to provide an individualized HEP to youths living with a rare condition.

To date, little is known about the potential effectiveness of exercises in adolescents with AMC [5]. Using this type of technology in a study that includes frequent follow ups as proposed in this protocol has the potential to reach more participants because it transcends geographical barriers, improving the sample size needed for research with heterogeneous populations such as AMC. Telerehabilitation allows inclusion of youths from across Canada, regardless of proximity to a specialized health care center. Although telerehabilitation has previously been used in studies on children with asthma and autism spectrum disorders [26,27], the only musculoskeletal disorder in which a telerehabilitation intervention was used is with children with cerebral palsy [18]. However, the results were variable among those studies which justify studies with other populations such as AMC [28,29]. In addition, little is known about the cost efficiency of this type of intervention [26,30].

Limitations

This pilot study has some limitations. Because of the remoteness of the assessment, even if passive range of motion is normally measured in the clinic, it will not be assessed in this study. Despite passive range of motion being important to measure, it is expected that therapists will have sufficient information to develop the individualized HEP from the initial telerehabilitation assessment. Another consideration is that youths will be instructed to wear the activity monitor on their wrists, even in the presence of upper extremity contractures that may interfere with the movement capture, therefore with the accuracy of step count. As the purpose of the activity monitor in this study is to measure heart rate and duration of the exercise sessions, and not to record steps, all youths will wear the watch at the wrist. Importantly, the goal of using telerehabilitation is not to replace face-to-face clinic visits or in-person therapy, but rather to propose a complementary intervention therapists can offer youths and their families during adolescence, at which time rehabilitation has been shown to decrease [12]. The aim to provide an individualized HEP is to help youths maintain their physical gains and assist them in reaching new goals that may arise as they grow. This telerehabilitation pilot study will also inform the possible pitfalls, beneficial effects, and cost associated with this new method of care, thus informing its use in other populations with musculoskeletal conditions.

Conclusion

Establishing the feasibility of using telerehabilitation for children with AMC will inform us for a future clinical trial. Information about the potential effectiveness of using telerehabilitation to deliver a HEP for children with AMC will be provided, thus leading to the development of a novel approach in this population. If proven successful, this service delivery model can be tailored to other pediatric musculoskeletal conditions, such as osteogenesis imperfecta and juvenile rheumatoid arthritis.

Acknowledgments

This study was awarded a Can $10,000 (US $7234.03) pilot grant from the Canadian MSK Rehab Research Network (CIHR FRN: CFI-148081) in May 2018. NDO received salary support from the Fonds de recherche du Québec—Santé.

Authors' Contributions

NDO initiated, planned, and received funding for this study. NDO wrote the study protocol. MG wrote the paper. JC, CE, GMM, JM, BS, RY, RH, LNV, and NDO revised the manuscript. MG, JC, CE, GMM, and RY are involved in the data collection.

Conflicts of Interest

None declared.

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Abbreviations

AMC: arthrogryposis multiplex congenita

APPT: Adolescent and Pediatric Pain Tool

AROM: Active range of motion

GAS: Goal Attainment Scale

HEP: Home-based exercise program

PAQ-A: Physical Activity Questionnaire for Adolescents

PODCI: Pediatrics Outcomes Data Collection Instrument

SHC-C: Shriners Hospital for Children - Canada

Chapter 4: Results

Connecting text

The protocol of the pilot study on the feasibility and effectiveness on the use of telerehabilitation was presented in chapter 3. In addition to the feasibility and effectiveness objectives set at the beginning of this project, the reliability of using a virtual goniometer with youths with AMC was assessed. The findings from this pilot study are presented in this chapter.

Manuscript 3: Feasibility and effectiveness of using telerehabilitation to deliver a home exercise program to youths with arthrogryposis: A pilot study

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This manuscript has been submitted to *International Journal of Environmental Research and Public Health* for the special issue “Evaluating Telerehabilitation for Patients across the Lifespan.” The manuscript is currently under review.

Abstract: Research on the use of telerehabilitation in children with musculoskeletal disorders is scarce. This pilot study aimed to evaluate the feasibility and to explore the effectiveness of a Home Exercise Program (HEP) delivered using telerehabilitation for youths with arthrogryposis multiplex congenita (AMC). A 12-week individualized HEP was developed using the Goal Attainment Scale (GAS) and measures of function (PODCI), physical activity (PAQ-A) and pain (APPT), and delivered remotely. Feasibility was measured using recruitment rates, compliance to the HEP, technical issues and assessing the reliability of using a virtual goniometer for range of motion (ROM) measurements. Effectiveness was explored using the Wilcoxon-signed rank test comparing pre-and post-results on ROM, PODCI, PAQ-A and APPT. The Goal Attainment Scale

(GAS) was used to assess if goals were achieved. Seven youths (median=16.9 years) completed the HEP. All feasibility criteria were reached except recruitment rates. A general ICC of 0.985 (95% CI: 0.980; 0.989) was found for virtual ROM measurement. Twelve of the fifteen goals set with participants were achieved. Statistically significant improvements were observed in the pain domain of the PODCI (p=0.08) and PAQ-A (p=0.03). Remote delivery of a HEP for youths with AMC was feasible. Promising results were found for the effectiveness of the HEP, but should be assessed with a larger sample.

Keywords: telerehabilitation; teleassessment; arthrogryposis multiplex congenital; physical therapy; occupational therapy

1. Introduction

Musculoskeletal (MSK) disorders affect 1 in 8 children and include injuries, disorders as well as congenital anomalies affecting both the spine and the extremities (1). MSK disorders are typically associated with pain and limitations in mobility and functional ability (2). Rehabilitation for individuals with acquired MSK disorders aims to recover from a loss of function; whereas, rehabilitation goals for individuals who were born with a MSK disorder change throughout the lifespan according to the individual's needs (3). For those with a congenital MSK disorder, the aim of early intervention is to reach motor milestones and maximize functional gains. Later in life, the focus of these goals shifts towards maintaining gains and maximizing function to enable participation in activities, school and later, employment (3).

Premature declines in function among adults with early onset MSK disorders have been reported in cerebral palsy, juvenile rheumatoid arthritis and osteogenesis imperfecta and may be attributed to muscle weakness, orthopaedic abnormalities, chronic pain and fatigue (4). These premature declines can lead to a sedentary lifestyle associated with a higher risk of secondary conditions (e.g., obesity, premature sarcopenia) which may in turn lead to cardiometabolic disorders, fragility and/or early mortality. This cascade can be prevented with adequate exercise. Indeed, exercise has been shown to increase muscle strength and prevent muscle atrophy, decrease pain and fatigue, and improve quality of life among youth with osteogenesis imperfecta (5), youth with juvenile rheumatoid arthritis (6) and adults with cerebral palsy (7).

Clinicians and researchers face challenges in developing adjunct therapies such as exercise programs for people with MSK disorders. Patients and their families, as well as therapists, report lack of time to physically get to the center and transportation to rehabilitation centers as barriers to exercise intervention (8, 9). These challenges can be further compounded with youths with rare MSK disorders as they often require specialized care. A rare disorder is characterized by its low prevalence in the population and is often defined as "a disorder that affects less than 1 in 2000 people in a population" (10). Given the rarity, many individuals live far from sub-specialized healthcare centers, thus limiting the provision of regular interventions with frequent follow-ups, such as exercise programs. To overcome such challenges, novel intervention approaches and technologies are needed to increase access to sub-specialized care for individuals living in remote areas. New approaches may contribute to reduced costs and decreased in-person visits to the hospital (11). Telerehabilitation, an innovative way to deliver rehabilitation services remotely using telecommunication technologies, may offer a solution to deliver intervention programs with frequent follow-ups (12). Despite its benefits, telerehabilitation has been understudied in children with physical disabilities (13).

One of these rare MSK disorders that may benefit from an exercise intervention through telerehabilitation is arthrogryposis multiplex congenita (AMC). AMC is an umbrella term used to describe over 400 conditions characterized by congenital joint contractures present in at least two body areas and affecting 1 in 4300 to 5100 live births (14, 15). Amyoplasia and distal arthrogryposis are the most common types of AMC and represent about 50% to 65% of all AMC diagnoses (16, 17). Individuals with AMC may have poor muscle mass in addition to their limitations in range of motion (ROM) resulting in difficulties in transfers, mobility and independent activities of daily living (18-20). One aspect contributing to the limited ROM in AMC is the lack of movement of limbs which may be overcome by physical exercise (18). Some studies speculated that exercise in youths with AMC may maintain ROM, increase muscle strength and decrease pain, but direct effects of an exercise program remains to be tested in this population (16, 18, 21, 22). A positive association was also reported between knee and hip muscle strength and motor function in AMC suggesting the importance of preserving sufficient muscle strength to maintain motor function (23). Currently, most interventions in AMC, specifically rehabilitation, occur in early childhood and frequency decreases during school-age and adolescent years, despite new challenges arising during these transition periods (24, 25). Rehabilitation for school-aged children focuses mostly on body functions and structure, which does not always correspond to the youth's specific needs, such as participating in activities and increasing independence for school and employment (3). The aim of this pilot study was to evaluate the feasibility of using telerehabilitation to provide a Home Exercise Program (HEP) for youths with AMC and to explore the effectiveness of this type of intervention. As part of the feasibility objective, the reliability of using a virtual goniometer for ROM measurements was assessed.

2. Materials and Methods

This prospective interventional single cohort pilot study took place between January 2019 and March 2020 at the Shriners Hospitals for Children (SHC) - Canada and received ethics approval from the McGill University Faculty of Medicine Institutional Review Board (#A08-B38-18B). Patients were included if they were between 8 and 21 years of age, understood written and spoken English or French, and had multiple congenital contractures as documented in their electronic medical records. Individuals were excluded if they had a recent surgery (i.e., 3 months for soft tissue and 6 months for bony surgery), lived outside of Canada or had cognitive deficits. Potential participants were identified by reviewing onsite medical records and were approached during their clinic visit, by postal mail or phone if they did not have an upcoming appointment. Recruitment was also sought by posting an ad on a Canadian AMC support group on social media. Informed consent forms were filled by all parents and youths 14 years and over. Assent was provided to youth from 8 to 13 years.

Intervention

All telerehabilitation meetings were provided remotely using Zoom pro (Zoom Video Communications Inc., United-States), a video-conferencing platform allowing an encrypted connection and synchronous exchange between participants and therapists.

Assessment

To establish the 12-week HEP, an initial assessment was conducted using a video-conferencing platform by an occupational therapist (OT) and a physical therapist (PT). This assessment included active ROM measurements of the upper and lower limbs, an evaluation of the overall function, and a pain assessment using the Adolescent and Pediatric Pain Tool (APPT). Based on the assessment, therapists and the participant determined individualized goals using the Goal Attainment Scale (GAS). The information obtained through this initial assessment was used by the rehabilitation team to develop an individualized 12-week HEP. In addition, participants were asked to complete an online questionnaire including the Physical Activity Questionnaire for

Adolescents (PAQ-A), the Pediatrics Outcomes Data Collection Instrument (PODCI), and baseline disorder and sociodemographic information (i.e., type of AMC, location of joint contractures, services care received, schooling, employment, volunteering, leisure).

At the end of the 12-week HEP, participants underwent a final assessment with the OT and PT using the same outcome measures as the initial assessment and completed the same online questionnaire with the addition of 19 closed-ended and 4 open-ended questions on their satisfaction with the HEP. The methods for the pilot study are described with more detail and can be found in Gagnon et al. (2020) (26).

Home Exercise Program (HEP) protocol

All information gathered during the initial assessment was used to build an individualized HEP for each participant based on their goals and their abilities. Therapists used Physiotec (Physiotec Quebec Inc., Canada), a software that easily builds HEP and provides participants access to detailed instructions and video of the exercises. Examples of these HEP can be found in the supplementary materials. A week after the initial assessment, a physical rehabilitation therapist (PRT), a professional whose role is to develop treatment plans and provide appropriate intervention in collaboration with a physical therapist, explained the HEP to the participant during a remote session. During this session, the PRT ensured that the participant understood and performed the exercises safely. Participants were asked to perform their HEP three times a week for approximately 15-30 minutes each time. Follow-ups were provided every three weeks (i.e. at weeks 3, 6 and 9) by the PRT to ensure exercises were well performed and to assess the level of difficulty of the HEP. Progression and/or regressions were offered accordingly. When necessary for optimal loading, materials such as resistant elastic bands or theraputty (a silicon-based exercise material used for hand therapy) were sent to participants by postal mail. In order to record the compliance to the HEP, participants were asked to wear a Polar watch A370 (Polar Electro Inc., Canada) to capture their exercise sessions which was sent to participants by postal mail and to complete a data-log sheet. The activity monitor was worn at the wrist during HEP and was used to capture the duration of the exercise sessions. Figure 1 illustrates a summary of the intervention.

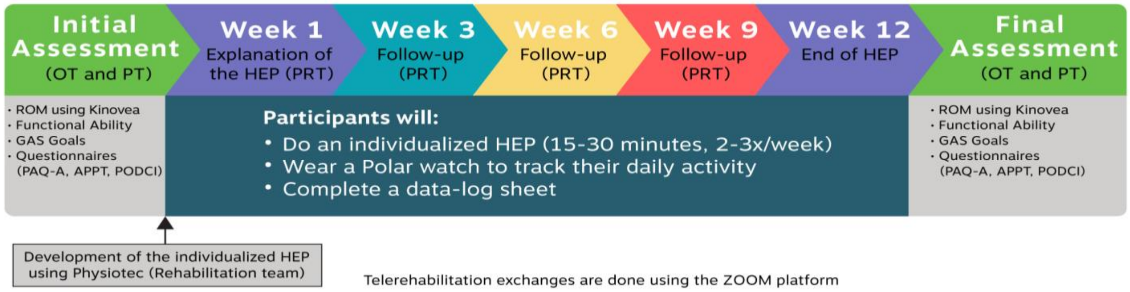

Figure 1. Timeline of the telerehabilitation intervention (26).

APPT: Adolescent and Pediatric Pain Tool; GAS: Goal Attainment Scale; HEP: Home-based exercise program; OT: Occupational therapist; PAQ-A: Physical Activity Questionnaire for Adolescents; PODCI: Pediatrics Outcomes Data Collection Instrument; PRT: Physical rehabilitation therapist; PT: Physical therapist.

Statistical Analyses

Summary statistics including the median and 95% confidence intervals (CI) for continuous variables and counts and proportions for categorical variables were produced for all variables. Given that this was a pilot study in a rare disease, statistical comparisons were produced only for exploratory purposes using a significance (α) level set a priori to 10% as it is proposed for research with rare disease (27). Statistical analyses were performed using IBM SPSS Statistics version 24 for Windows (IBM Corp., Armonk, NY, USA).

Feasibility

Feasibility was evaluated through different operationalization criteria (i.e, recruitment rates, withdrawal rates before and during the intervention, completion rates, compliance to the HEP and to the telerehabilitation meetings, % of missing data, technical issues). These operationalization criteria are defined in detail in the published protocol for this study [26]. Compliance to the HEP and the telerehabilitation meetings were described using summary statistics and were compared to pre-established feasibility criteria using one sample Wilcoxon signed rank test. For the remaining operationalization criteria comparison with pre-established criteria was done using the same method. Participants' experience regarding use of technology and overall satisfaction with the program (assessed with four open- and 19 closed-ended questions), and technical issues (major technical issues being an issue that resulted in the cancellation of the meeting and, minor issues being something that can be resolved during the meeting) experienced during the program were also descriptively assessed with summary statistics.

The inter-rater reliability of using a virtual goniometer to measure the active ROM has been shown to be feasible on ten healthy adults (28), but not in children and adolescents with a MSK disorder such as AMC. The inter-rater reliability was established by having two raters (MG, GMM) measure the ROM of four participants selected at random using a virtual goniometer (Kinovea, version 0.8.15). The intraclass correlation coefficient (ICC) and associated 95% CI for each joint and overall were calculated based on single measurement, absolute-agreement, 2-way random-effects model (29-31).

Effectiveness

The effectiveness of the HEP was assessed using the GAS. Raw GAS scores were converted to GAS T-scores for each participant and for the global HEP (32). Within-participant pre- and post-intervention scores of the PODCI, PAQ-A, APPT and ROM were compared using the related-samples Wilcoxon signed rank test. Individual changes were also compared to evaluate clinically important differences as defined by Oeffinger et al. (33) in the following domains of the PODCI (changes in points associated to a minimum clinically important difference): upper extremity (4.8 points), transfers and mobility (5.2 points), sports and physical function (10.3 points), pain and comfort (26.3 points), happiness (11.2 points) and global function (8.2 points). Changes in the APPT for the 10-points pain intensity scale were considered clinically meaningful when there was a change greater than two points (34, 35).

For the questionnaires, a sensitivity analysis was conducted due to a participant having suffered from an ankle injury, unrelated to the HEP execution, before the final assessment. An imputation technique was used to replace final results of the participant that were directly affected by the injury. Final results were replaced by the median of the group in the PAQ-A, and in the following domains of the PODCI: transfer and mobility, sport and physical function, pain and comfort and global function to perform the Wilcoxon signed rank test.

3. Results

3.1. Participant information

Of the 114 patients with AMC follow at SHC-Canada, 11 consented to the study, but five boys and two girls completed the intervention. Participants were recruited in clinic (n=6), by postal mail (n=2), by phone (n=2) or on social media (n=1). Of the four participants who withdrew, two consented in clinic, but were not reachable to schedule the initial assessment, one withdrew due to personal reasons and one withdrew as the caregiver could not supervise the HEP. See Figure 2 for participation flow diagram. Participants' median age was 16.9 years old (range: 11.3 to 20.8 years old) and represented four Canadian provinces with a median distance from the SHC-Canada of 227 km (range: 7 km to 3439 km). Two participants had amyoplasia, four had distal AMC and one had an unknown type of AMC. Joint involvement was distributed as follows: shoulders (n=5), elbows (n=6), wrists (n=6), hands (n=4), hips (n=3), knees (n=5), ankle (n=6) and spine (n=1) with a median of six (95% CI: 3 to 8) joints involved by participant. For the level of ambulation, four participants were classified as community ambulators, two as household ambulators and one as non-ambulatory.

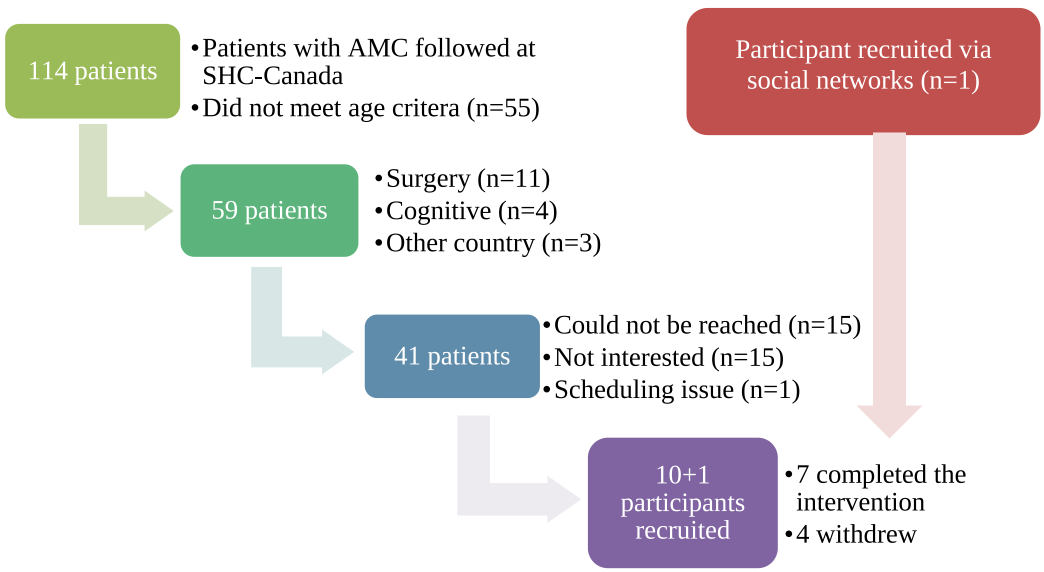

Figure 2. Participants' flowchart

3.2 Feasibility

3.2.1 Operationalization criteria

Recruitment rate among eligible participants who were reachable was 10/26 (38.5%) which was inferior to the target set at ≥ 50%. The withdrawal rate before the start of the intervention was 2/11 (18.2%), and 2/11 (18.2%) after the start of the intervention which met the established criteria of ≤ 20% and ≤ 30%, respectively. Completion rate of the HEP was 7/11 (63.6%) which corresponds to the criteria of ≥ 50%. The number of meetings that needed to be rescheduled in less than 24h notice was 5/47 (10.6%) which reached the target of ≤ 15%. The median HEP compliance was 2.09x/week (95% CI: 1.25 to 4.08x/week), which when compared to the three weekly sessions set in the HEP, corresponds to 69.4%, and is higher than the target of ≥50% (p=0.046). Different technical issues arose during the telerehabilitation meetings. No major technical issue necessitating cancelling a meeting occurred, but minor technical problem happened in 5 of the 47 meetings (10.6%). Echo voices were present with one participant and this was resolved with the muted option so that when the therapists or the participant spoke, the others muted themselves. The therapist lost the connection in one meeting for a few seconds and had to reconnect. On one occasion, a participant had a difficulty with the sound of their computer and had to join with their phone as an alternative. For material issues, five participants had difficulties syncing the watch and the project coordinator resolved this problem by sharing the screen during the telerehabilitation meetings to review the required steps for setting up the watch.

All participants completed the pre- and post-intervention questionnaires except for one participant who did not return back the data log-sheet and never synced their watch, so they were excluded from the HEP compliance calculation despite reporting having performed their HEP regularly. Another participant did not wear the watch. For the ROM measurements, missing values for each joint can be found in the supplementary table.

3.2.2. Inter-rater reliability

Inter-rater reliability varied among different joints with an ICC of 0.985 (95% CI:0.980; 0.989) for all joint combined. The lowest ICC being the forearm pronation with a median ICC of 0.252 (95% CI: -0.477; 0.75) and the highest being the shoulder extension with a median ICC of 0.998 (95% CI: 0.983; 1.00). Specific ICCs for each joint can be found in table 1.

Table 1. Intraclass correlation for each joint

Joint	Number[a]	Median	95% CI
Shoulder abduction	10	0.668	(0.072, 0.908)
Shoulder flexion	4	0.915	(0.158, 0.994)
Shoulder extension	5	0.998	(0.983, 1)
Elbow flexion	12	0.988	(0.956, 0.997)
Elbow extension	12	0.99	(0.965, 0.997)
Forearm pronation	10	0.252	(-0.477, 0.75)
Forearm supination	10	0.691	(0.069, 0.917)
Wrist flexion	12	0.707	(0.277, 0.904)
Wrist extension	12	0.858	(0.073, 0.968)
Hip flexion	14	0.72	(0.315, 0.901)
Hip extension	11	0.833	(0.484, 0.952)
Hip internal rotation	11	0.975	(0.914, 0.993)
Hip external rotation	12	0.971	(0.901, 0.991)
Knee flexion	14	0.992	(0.958, 0.998)
Knee extension	16	0.986	(0.961, 0.995)
Ankle dorsiflexion	12	0.917	(0.749, 0.975)
Ankle plantarflexion	12	0.878	(0.636, 0.963)

1. Number of data included for analysis with a possible maximum of 16 (4 participants * 2 sides).

3.3. Effectiveness

For the GAS, objectives varied among participants and related to pain management; endurance in writing, standing or walking; sports (sledge hockey and karate); daily activities (buttoning up shirt, transfer ability, control stair descent, self-propelling the wheelchair) and strengthening. The median number of goals established with the participants was 2 (range: 1 to 3). A total of 15 goals were established and 12 goals were achieved. The overall program t-score was 74.85 whereas the median t-score among participants was 56.21 (30 to 72.82).

Pre-and post-intervention results from the different questionnaires can be found in table 2. A statistically significant change was observed in the pain and comfort domain of the PODCI (p= 0.08). The number of participants with clinically important changes were as follows: upper extremity (improvement, n=2; decrease, n=1), transfers and mobility (improvement, n=3; decrease, n=2), sports and physical function (improvement, n=1; decrease, n=1), pain and comfort (improvement, n=1; decrease, n=0), happiness (improvement, n=2; decrease, n=1) and global function (improvement, n=2; decrease, n=0). For the PAQ-A, a statistically significant important improvement of 24.6% was observed for the group (p=0.03). For the APPT, four of seven participants reported pain at baseline. No changes were statistically significant in the above-mentioned questionnaire, but one had a meaningful clinically improvement.

Table 2. Pre-and post-results from the different questionnaires

Questionnaires		Pre-intervention		Post-intervention		Significance (p value)
		Median	95% CI	Median	95% CI	
PODCI[a]	Upper extremity	87.50	(12.50, 100)	95.83	(4.17, 100)	0.79
	Transfer & mobility	93.94	(21.97, 100)	91.66	(12.12, 100)	0.60
	Sport and physical function	79.86	(11.36, 86.11)	63.64	(13.64, 93.18)	0.61
	Pain and comfort	71.11	(34.44, 100)	85.00	(49.44, 100)	0.08*
	Happiness	85.00	(50.00, 100)	90.00	(35.00, 100)	0.50
	Global function	87.47	(20.07, 94.86)	82.08	(19.84, 94.55)	0.61
PAQ-A[b]		1.09	(1.00, 1.41)	2.32	(1.00, 3.45)	0.03*
APPT[c]	Location	1.00	(0.00, 6.00)	1.00	(0.00, 6.00)	1.00
	Scale (cm)	1.55	(0.00, 5.50)	1.10	(0.00, 6.20)	0.72
	Sensory (%)	5.40	(0.00, 10.8)	2.70	(0.00, 10.8)	> 0.99
	Affective (%)	0.00	(0.00, 0.00)	0.00	(0.00, 0.00)	> 0.99
	Evaluative (%)	0.00	(0.00, 25.00)	0.00	(0.00, 37.5)	> 0.99

* Statistically significant (p <0.1)

a. Pediatric Outcomes Data Collection Instrument

b. Physical Activity Questionnaire for Adolescents

c. Adolescent Pediatric Pain Tool

For the ROM, improvements for shoulder abduction (p=0.08), shoulder flexion (p=0.07), wrist extension (p= 0.05) and knee extension (p=0.04) were found to be statistically significant. The other joints did not show significant changes. The results for each joint ROM can be found in table 3.

Table 3. Range of motion

Joint	N[a]	Pre-intervention		Post-intervention		Significance (p value)
		Median	95% CI	Median	95% CI	
Shoulder abduction	12	143	(16, 159)	151.5	(25, 163)	0.08*
Shoulder flexion	6	34.5	(0, 152)	57.5	(0, 153)	0.07*
Shoulder extension	3	66	(66, 75)	60	(50, 66)	0.11
Elbow flexion	14	146.5	(86, 164)	144.5	(92, 158)	0.67
Elbow extension	11	(-9) [b]	(-82, 0) [b]	(-4) [b]	(-95, 9) [b]	0.35
Forearm pronation	9	82	(65, 91)	95	(78, 102)	0.12
Forearm supination	9	71	(44, 86)	66	(49, 84)	0.48
Wrist flexion	8	86.5	(29, 97)	78	(11, 94)	0.11
Wrist extension	8	20.5	(-20, 50) [b]	30	(-7, 57) [b]	0.05*
Hip flexion	12	109.5	(95, 127)	108.5	(94, 130)	0.92
Hip extension	8	1.5	(-50, 30) [b]	(-1.5) [b]	(-45, 16) [b]	0.25
Hip internal rotation	3	1	(1, 27)	12	(10, 27)	0.18
Hip external rotation	4	30	(4, 55)	33	(11, 48)	0.72
Knee flexion	12	105.5	(50, 130)	115	(93, 138)	0.61
Knee extension	14	(-18) [b]	(-63, -7) [b]	(-14.5) [b]	(-62, -4) [b]	0.04*
Ankle dorsiflexion	12	(-21) [b]	(-28, 5) [b]	(-20.5) [b]	(-29, -2) [b]	0.72
Ankle plantarflexion	12	32	(23, 40)	29.5	(21, 41)	0.53

* Statistically significant ($p < 0.1$)

a. N=number of data included for analysis with a possible maximum of 14 (7 participants * 2 sides).

b. The negative value represents lack of ROM. For example, negative knee extension signifies inability to achieve full extension.

3.4. Satisfaction

All seven participants reported being comfortable communicating through the telerehabilitation platform, and that the clarity of the video and audio of the platform was acceptable. Participants also reported reduced travel time compared to if they had to come in-person. Participants felt supported during the 12-week program and expressed interest in using telerehabilitation in the future. All were satisfied or very satisfied with the remote evaluation, delivery of the HEP, follow-ups and the overall organization of the 12-week program. In the open-ended questions, participants mentioned feeling content with the personal improvements made (n=4), support from the staff (n=3), the motivation provided by the intervention to do their exercises (n=3) and the method of intervention (n=3). When asked to identify constructive feedback for future programs as well as recommendations and possible solutions, suggestions were made to provide information about the possibility of using a phone application to sync the watch in addition to using the computer (n=1), use a more trustworthy telecommunication platform (n=1), increase the span of the program (n=1), include an in-person follow-up visit halfway through the program (n=1), and include outcome measures targeting the adolescent age group rather than younger children (n=1). Also, participants reported having more strength (n=3), endurance (n=2), mobility (n=1) and balance (n=1) at the end of the 12-week HEP as compared to before. In addition, two participants noticed a positive change in their motivation to complete their program, and three reported improvements in their daily activities including transfers, descending stairs, walking ability and greater efficiency propelling a manual wheelchair.

4. Discussion

In this study on the use of telerehabilitation to deliver a HEP to youths with AMC, it was found that this approach is feasible and well accepted by participants. The reproducibility of using a virtual goniometer was found to be good to excellent except for the forearm pronation ROM. As the main objective was to assess the feasibility, this pilot study also aimed at determining the most suitable outcome measures for this type of intervention. It was found that the GAS corresponded better to the need of a telerehabilitation intervention and to the reality of having a heterogenous population with AMC. The effectiveness of the intervention was explored and most of the individualized goals were achieved and significant improvements were observed in the pain domain of the PODCI, the PAQ-A, and various ROM.

4.1 Feasibility

The feasibility has been demonstrated with the achievement of all operationalization criteria except for the recruitment rates. Although the efficiency of different recruitment methods was not directly studied, it was noticed that it was easier to recruit participants directly during their clinic visits instead of postal mail or phone. However, some of the older individuals with AMC are followed only as needed; therefore in-clinic recruitment would not be suitable to recruit them. The observed completion rates of 63.6% correspond to the criteria set at being ≥ 50%. One withdrawal was due to the need for more support from the caregiver to do the HEP. The support from families or the entourage of the participant was important for most of the participants, specifically for the younger participants and those more severely affected by AMC. In most of the telerehabilitation meetings, a parent was present to help position the camera, position or assist the participant or aid in the exercise material set-up. In one case, the telerehabilitation meetings took place at the participant's school with the assistance of school therapist. The proposed intervention in this study was semi-supervised with follow-ups provided every three weeks. The compliance of 69.4% to the HEP measured in this study is closer to the compliance rates of 76% observed in supervised interventions (36) than to that of non-supervised interventions (11-37%) (37). A total of 11% of the telerehabilitation meetings were cancelled on the same day which is similar to the 12% observed for in-person appointments within the rehabilitation department at SHC-Canada and the rate of missed appointments of 12% observed in an adolescent unit at an university hospital in Switzerland (38). Another observation is that hospital appointments are often cancelled during extreme weather conditions in Canada, but no appointment was cancelled for this reason in this study due to the remote aspect of care delivery. A few technical issues occurred during this study, but all were resolved promptly and none affected the safety of the participants. Therefore, technical issues should not be a reason to preclude the use of telerehabilitation in future studies or in clinical practice.

The custom close-ended question survey used showed that the satisfaction with the telerehabilitation approach was high among participants. However, it could be useful in order to interpret the results and compared to other studies to use a standardized questionnaire such as the Telehealth Usability Questionnaire (39), but none seem to be validated specifically for adolescents. All participants agreed or strongly agreed that the quality and clarity of the video and audio of the telerehabilitation platform was acceptable. Yet, in the closed-ended questions, one participant mentioned that improvements could be made for future similar interventions.

All selected questionnaires for the baseline and final assessments were validated for children up to 18 years, which corresponded to the majority of the participants included in this study (5/7 participants). However, at SHC-Canada, patients are followed until 21 years of age, and one participant in this situation mentioned the content of some of the questionnaires were not adapted to their reality. Finding standardized measures and questionnaires devised and validated for both, pediatric and young adult populations, is often a challenge when they are grouped within the same study.

Feasibility of virtually measuring ROM posed some challenges. Normally, during an in-person assessment, therapists instinctively select the adequate position to perform accurate ROM measurement. However, when done virtually, challenges were faced in that some ROM measurements were not always done in the correct movement planes. In order to ensure complete data capture when remotely assessing ROM, we recommend the followings: developing guidelines to ensure movements are performed in the proper plane of movement in front of the camera;

training the therapists with the guidelines prior to the beginning of the telerehabilitation intervention and completing a form. However, despite the goodwill of the therapists, the environment of the participant sometimes limited the possibility of having a perfect screenshot of ROM due to space restrictions/limitations in the home, or poor or too high luminosity. Different strategies can be used to enhance ROM data capture: (1) Asking participants to wear clothes contrasting with their background; and (2) using a laptop or a tablet allowing moving the camera in the proper position to measure the ROM in the required plane of movement. Although reproducibility was not optimal, it was nevertheless sufficient to set the HEP goals and complete training sessions.

Reliability was shown to be higher and less variable for lower extremity joints than for upper extremity joints. Selecting participants randomly, led to the inclusion of a participant with no upper limb active ROM in the analysis thereby limiting the number of data to assess ICC for the upper extremity. The low sample of data for upper extremity may have impacted the reproducibility of upper limbs ROM. Furthermore, participants had less possibility for compensatory movements for their lower limb joints, which facilitated measurement-taking. Lower limb joints ROM were taken lying on the floor while upper limb measurements were taken with participants standing or sitting on a chair with more possibility to compensate. The moderate to good reproducibility observed in this study suggest that virtual assessment of ROM is feasible. However, raters used the same screenshots, which may have improved the reliability as we removed a source of potential bias. Nevertheless, it was not the scope of this study, a test-retest of the ROM measurements using a virtual goniometer and a validation study showing good agreement between in-person and virtual ROM would strengthen this position.

4.2 Effectiveness

Results about the effectiveness are promising. The GAS, which was the main outcome for effectiveness, showed significant improvement at the end of the 12-week HEP. Among the 12 goals achieved using the GAS, nine goals were achieved beyond expectations. The evaluation of the goals was dependent of the goals, for example increasing the number of words written in certain time, percentage of perceived strength, scale from 0-10 of satisfaction or walking endurance in minutes. These large improvements might be due to how goals were scaled using the GAS table. As most of the participants did not do exercises prior to participating in this project they may have demonstrated quicker gains. However, two participants set a goal related to pain and both did not achieve this goal perhaps due to the multifactorial etiology of pain requiring management by an interdisciplinary team (40). Also, both participants had chronic pain for a long period of time when the goal was set, and it may not be realistic to expect that a 12-week HEP would lead to significant pain reduction. Apart from the pain goal, the other goal that was not achieved was concerning walking endurance. According to the participant herself, the baseline walking endurance level may have been overestimated. Therefore, despite improvements reported by the participant at the end of the 12-week HEP (quicker walking pace and being able to follow the class during a 3-week field school trip), significant improvement was not attained. As the GAS allows for individualized and varied goals, it proved to be a versatile and sensitive measurement tool, given the high level of variability of the participants joints involvement and physical function. Despite the benefits of using the GAS, it is important to know that this tool requires some level of training and experience to develop goals that are participant specific, measurable, achievable, realistic and timed (SMART) (41). After having determined individualized goals in collaboration with the participants, therapists need to scale the goal according to an expected level of change, and predict potential improvement that will be achieved within the 12-weeks after only one initial evaluation with the participant, which may not always be intuitive.

In contrast to the GAS, PODCI showed a significant improvement only in the comfort and pain domain. A potential reason for that observation is that participants had set specific goals related to one specific domain of the PODCI. The improvements were thus lost amongst the other domains of the PODCI showing no improvement or even a decline. Also, a floor and ceiling effect were noticed in a few participants. For example, for the participant with the lowest physical function level, all but the happiness domain demonstrated a floor effect. Therapists noticed a large improvement in the transfer ability from wheelchair to exercise table in a non-ambulatory participant, yet the PODCI did not detect this change since the PODCI focus mainly on ambulatory activities such as walking and running. The opposite effect also occurred in participants that scored maximum scores at baseline and therefore no positive change was detected. For these reasons, we conclude that the PODCI may not be the appropriate measure for our study using an exercise intervention in a heterogenous group of youths.

Baseline results from the PAQ-A were very low yet increased at the end of the 12-week HEP with levels approaching those of typically developing adolescents (42). The level of physical activity among participants with AMC remained lower than typically developing adolescents similarly to a study with adolescents with CP (43). However, the PAQ-A is not adapted for individuals with MSK disorders such as AMC, and most of the exercises proposed in this study were for the most part, rehabilitation exercises which are not measured by the PAQ-A (focus on physical activity). Although several participants reported participating in adapted physical activity, the PAQ-A does not provide such a response option.

At final, based on participants' individual objectives we did not expect any significant changes in ROM as no participants selected this as a goal. However, few statistically significant improvements were found and may be explained by an improvement in muscle strength which may indirectly lead to an increase active ROM. These ROM results should be taken with caution as changes may also be due to measurement errors.

In the satisfaction questionnaire, five participants mentioned having improved their goals set using the GAS, and five participants reported additional improvements that were not included in their initial goals. One participant wanted to reduce the pain level which was not improved during the intervention, but the participant mentioned having improved transfer ability. Another participant had goals related to endurance, which were achieved, but also reported having improved his overall strength. Another participant aimed to improve walking endurance and despite not achieving this goal, noted improvement in balance and muscle strength. Two participants reported increase motivation in regards to regular exercise.

4.3 Limitations

This pilot study presents with some limitations. Passive ROM, strength and function are normally assessed in clinic, but due to the remoteness of the assessment, it was not feasible to do so in this study. However, therapists had enough information with the active ROM, the GAS and the information provided by participants during the initial assessment to build an individualized HEP.

The small sample size, coupled with withdrawal rates and the heterogeneous population, makes it difficult to generalize the findings about the effectiveness of the intervention proposed in this study to a wider population of youths living with AMC. In addition, most of the outcomes were standardized. Even if the HEP was based on individualized goals, assessments were not individualized and did not always correspond to the need of the participants. The assessment provided in this study did not give objective information related to the individualized goals. For example, joint-specific ROM was measured for all participants, but it was only pertinent for one participant who had a dressing goal. Most of the goals were related to muscle strength and endurance, but we did not assess objectively those outcomes and we relied solely on the subjective component of the evaluation.

5. Conclusions

The results of this pilot study suggest that it is feasible to use telerehabilitation to deliver a HEP for youths with AMC. Telerehabilitation is not only useful for people living remotely, but also for those living close when they need consultations during pandemic (such as covid-19) when social distancing is needed (44). This approach also holds promising results regarding the effectiveness of the HEP but remain limited given the small sample size. A larger multi-centered randomized control trial would be needed to ascertain an adequate study power to conclude on effectiveness. Finally, lessons can be learned from the challenges and positive aspects denoted in the current study, which could lead to enhance future protocols.

Supplementary Materials: The following are available online at www.mdpi.com/xxx/s1, Document S1: Examples of exercise programs, Table S1: Missing value for ROM measurements,

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Funding: This research was funded by the Canadian Musculoskeletal Rehab Research Network, (CIHR FRN: CFI-148081), Can $10,000. MG was awarded a Masters scholarship from the Grand Défi Pierre Lavoie Foundation, Can $7500. NDO received salary support from the Fonds de recherche du Québec-Santé.

Acknowledgments: We owe a special thanks to all youth and their families for their participation in this project. We acknowledge the contribution of Dr. Emmanouil Rampakakis for his advice on statistical analysis and his review of the manuscript. We thank Guylaine Bédard from the SHC-Canada for the illustration.

Conflicts of Interest: The authors declare no conflict of interest.

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Chapter 5: Discussion

The purpose of this masters project was to answer two questions that arose by key stakeholders (youths and adults with AMC, parents and clinicians) during an AMC knowledge exchange day held in May 2016. The scoping review proposed in this thesis helped to answer the question of what currently exists as assessments and interventions for physical function in children with AMC. Through this scoping review, we found that osteotomy was the most common surgery and rehabilitation the most frequent conservative intervention for children with AMC. Better quality studies are needed to determine what works best for children with AMC in order to establish evidence-based practice guidelines. One of the other priorities arose during this knowledge exchange day was about the effectiveness of exercise intervention for individuals living with AMC. With the scoping review, we found that little was known about the effect of exercise in AMC and decided to develop an exercise intervention. To be able to offer adjunct therapies to the population followed at SHC-Canada, we had to implement an innovative approach that would allow us to reach out to them without having them to come in person. Since most of our patients lives at least 100 km from the hospital, telerehabilitation was suggested as a potential solution. Telerehabilitation was known to be used with adults. This approach has been shown to be as effective as in-person intervention for individuals with total knee arthroplasty (9, 21) and stroke (22). Telerehabilitation is well accepted by patients and care providers (18, 23). Based on the positive results obtained in other populations, telerehabilitation was suggested. However, because there was a gap in the literature regarding using this approach in pediatric population with musculoskeletal disorders, a feasibility study was needed before to implement a large randomized control trial study (24). Evaluating the feasibility of using telerehabilitation for remote care delivery was therefore the second aim of this thesis. This project is the first step in order to determine if an exercise intervention can help individuals living with AMC. Our study showed that it was feasible to use telerehabilitation in youths with AMC. However, different aspects such as policies or the type of assessment or approach need to be taken in consideration in the development of a larger study or before implementing telerehabilitation in clinical settings.

Policy Issues in Regards to the Use of Telerehabilitation

In the past two decades, telerehabilitation started to be studied and used clinically, but professional practice and insurance policies are not yet perfectly established (25, 26). As a tertiary health-care center, SHC-Canada received local, provincial, national and international patients' referrals. Patients coming from everywhere raised some feasibility issues in this study not raised in previous studies that used telerehabilitation for local or provincial purpose. In our study, we had to add an exclusion criterion related on patients' location due to policies and regulations. Three eligible patients had to be excluded as they came from outside Canada. We needed to take a special licence for four participants coming outside Quebec province. Professional practice and insurance policies vary according to the location of clinicians and their respective professional orders. Because those policies vary across geographical regions and professions, every clinician interested in implementing this approach should be aware of the specificities of the policies in effect within their regions and professions. For example, physical therapists and physical rehabilitation therapists in the Province of Quebec are covered by their responsibility insurance to provide rehabilitation services in Canada. They nevertheless would have to account for the regulation in effect in the region where their patients will receive telerehabilitation cares (27). For occupational therapists, their provincial order currently does not mention the possibility of providing services outside the province (28). To be able to practice in other Canadian provinces, occupational therapists need to have a Canadian licence from the Canadian Association of Occupational Therapists. This license allows practicing anywhere in Canada and using telerehabilitation for care delivery (29). Position statements and guidelines are published by different orders and organizations such as the World Federation of Occupational Therapists (30, 31), the American Occupational Therapy Association (32) and the Canadian Alliance of Physiotherapy Regulators (33). All agree with the use of telerehabilitation, but mentioned the necessity of obtaining the consent from patients and, that clinicians should meet the same standards of care as services delivered in person. Another aspect that needs to be considered is private insurance coverage for private practice. Some insurance companies cover the fees associated with a telerehabilitation intervention, whereas others do not mention if this approach is covered. Therefore insurance coverage should be verified prior to provide telerehabilitation services (34). Also, governmental organisms such as the "Commission des normes, de l'équité, de la santé et de la sécurité du travail (CNESST)" or the "Société de l'assurance automobile du Québec (SAAQ)" took special measures during the covid-19 pandemic and allow the use of telerehabilitation to facilitate access to care. However, it is not known if telerehabilitation will still be accepted by those governmental organisms when the pandemic will be over (35, 36).

Clinical Implication

At the beginning of this project, we anticipated that telerehabilitation would be a useful approach to develop new interventions requiring frequent follow-ups for people living far from specialized hospitals. However, during the past few months, we learned that telerehabilitation is also an appropriate approach for service delivery during exceptional situations such as a pandemic (37). Recently, telerehabilitation became a necessary approach to use in the context of the COVID-19 pandemic situation to limit in-person visits and was implemented largely in daily clinical practice even for patients who live nearby the hospital, as well as in private practices. Telerehabilitation has gained a lot in importance, and research on the use of telerehabilitation increased in diverse populations and in different countries, including developing countries (38-40). Results of this thesis showed that using telerehabilitation to administer adjunct therapeutic interventions was feasible in a youth population with AMC. Despite being very specific to this population, one could hypothesize that it is probably feasible to use telerehabilitation in other paediatrics populations with orthopaedic conditions with similar needs as AMC such as osteogenesis imperfecta. However, in this project, patients with cognitive issues or who undergo recent surgery were excluded; therefore, before implementation in a clinical milieu, telerehabilitation should be tested with those populations.

In this project, we used telerehabilitation to deliver a home-based exercise program (HEP) with follow-ups every three weeks. This type of intervention led to a compliance to the HEP of 69.4% which is similar to the 76% observed in supervised intervention (14). However, the compliance is not the only aspect that needs to be considered to determine the best approach. Individuals may perform their exercises with full commitment if they are constantly supervised than if they are not. A study with stroke survivors compared a 10-week supervised and unsupervised exercise intervention. The study showed that both groups improved, but there was a tendency with better improvement for the supervised group and gender differences after one year (41).

In our intervention, an occupational therapist, a physical therapist and a physical rehabilitation therapist were involved. However, individuals with AMC could benefit from a more extended team working with an interdisciplinary approach. Interdisciplinary approach has been shown to be effective for pain and weight loss (42, 43). Depending on the goals of the youths, in addition to therapists involved in this project, kinesiologists, nutritionists, psychologists and pain physicians could be a great asset to maximize gains. For example, a child may want to improve his/her walking ability, but the presence of pain, being sedentary and overweight makes it more difficult to reach the goal. Therefore, an interdisciplinary intervention could be beneficial.

Teleassessment

In the current pilot study, we established that a remote ROM assessment with a virtual goniometer shows good reliability. This is useful for clinicians, but additional assessments are needed to evaluate the functional ability of individuals. For example, balance should be assessed in order to develop an exercise program. During the intervention, the physical rehabilitation therapists noticed that for some exercises, balance was an issue for participants and needed to be improved before being able to provide exercises that required a minimum of balance such as step down or lunge. The development of wearable devices or the adaptation of tests currently used in clinic could be a great addition to improve the quality of assessments performed from remote. Different tests that are currently used in clinic, necessitating little space and no direct contact with patients might be adapted and assessed by video-conferencing. For example, thirteen of the goals set in this project were functional, but no remote assessments were used to assess this aspect of the project. Functional assessments that should be feasible remotely with youths include for example the timed-up and go, the thirty-second sit-to-stand or the five-times-sit-to-stand tests. These tests have begun to be used for remote assessment in adult populations, but should be explored with adolescents (10, 44). In order to improve the quality of remote assessments for those people for who it is difficult to come to specialized health-care centers, wearable sensors that could be sent by mail have the potential to help obtain objective values. These sensors are currently used in different studies related to sports for injury prevention or coaching purposes (45, 46), and begin to be used in clinical settings. For example, inertial measurement units have the potential to be used to assess gait pattern, kinematic during different functional tasks, balance or wheelchair propulsion assessment performed at home (47-50). Surface electromyography or digital handheld dynamometer that could be connected to a computer by USB or Bluetooth and allowing therapists to see the results at the same time on the screen could be useful to assess muscle function, particularly in the context of an exercise intervention (10, 51). In the case of a physical activity intervention, physical activity monitors may be used to capture different outcomes such as heart rates, duration of the session, steps or intensity.

Knowledge translation

Knowledge translation (KT) methods are important to share information to other researchers, clinicians, individuals living with AMC and their families, and decision makers. KT methods help reach knowledge users quickly in order to decrease the gap between research and clinical practice (52). Results from this thesis will be shared using traditional KT dissemination strategies (publications in scientific journals and conferences) and non-traditional methods (video, infographic and podcast). Non-traditional methods will be used to share findings with other target audiences than researchers (individuals with AMC, families and clinicians).

In order to decrease the gap between research and clinical practice, this thesis was based on what knowledge users mentioned during the knowledge exchange day in 2016. This approach helps make sure that we answer a need in current clinical practice raised by individuals living with AMC, clinicians and other key stakeholders. In order to share findings to clinicians, a team at SHC-Canada currently develop rehabilitation guidelines based on the scoping review presented in this thesis and other scoping reviews made on AMC (53, 54). These guidelines will help to spread the information to clinicians that do not necessarily have the resources to conduct literature searches and appraise the evidence. Despite the promising results stemming from this project, telerehabilitation intervention is not ready to implement yet in clinic for an exercise intervention. The principal aim of this telerehabilitation pilot project was to assess the feasibility and to explore the effectiveness of an exercise intervention for youths with AMC. This approach needs to be studied among a wider group to determine the effectiveness of a HEP. To share knowledge on telerehabilitation to clinicians, direct consultations with those who want to implement telerehabilitation in their practice can be done. During the COVID-19 pandemic, we were approached by a physical therapy clinic that wanted to implement telerehabilitation in their practice. The consultation helped them with this approach to cope with the recommendations from the government stipulating that only essential face-to-face encounters were allowed initially.

To share the project to individuals with AMC and their families as well as to a lay audience, a video explaining the project was created and shared on social media[1]. An infographic and a

[1] This video was presented at the RSBO 2020 video competition and received the public prize (500$). The video has been viewed about 10 000 times on different social media platforms. https://www.facebook.com/1526980480924557/videos/276176690402136

podcast with the main outcomes from this project are in development and it is planned to be shared soon with individuals with AMC and their families on different social media platforms. The infographic can be found in the supplementary materials.

In addition to this plan of dissemination, we used traditional methods to reach researchers, decision makers and clinicians. Results from this thesis were shared through publications in scientific journals and presented at local, provincial and national scientific conferences and rehabilitation symposium. Additionally, the scoping review will be presented at the International Conference in Knowledge Translation in Rehabilitation and the telerehabilitation project will be presented at the 74th annual meeting of the American Academy for Cerebral Palsy and Developmental Medicine.

Scope and limitations

Sources of bias that are inherent in research studies include selection and information bias and are reported here in the context of my Masters study. Selection bias is the bias introduced by the selection of individuals which may negatively affect the statistical analysis. In my project, all patients with AMC followed at SHC-Canada and meeting the eligibility criteria were contacted for participation. The methods to invite potential participants to the study included telephone calls, in-clinic visits and mailed invitation letter. This method of selection reduced the possibility of selection bias as all eligible participants were invited, regardless of their physical functional level. However, the selection of individuals did not represent all individuals with AMC. Youths with cognitive impairments were excluded, even though about 23% of individuals with AMC has cognitive deficits (55). AMC included about 400 types (56), and the sample included in the telerehabilitation study mainly represents the two most common types of AMC which are amyoplasia and distal AMC (57).

The variability in our participants (e.g. geographical location, age and functional level) showed the low source of selection bias. Participants came from four Canadian provinces, were aged between 11.3 and 20.8 years old and has level of ambulation going from motorized wheelchair with assistance for self-care activities to independent walker. However, to ensure participant can take part in the intervention, participants with cognitive deficits were excluded in this feasibility study, but they should be included in the next phase as all youth with AMC may benefit from using a remote approach to deliver a home exercise program. Therefore, the feasibility of using telerehabilitation with youths with AMC having cognitive deficits is not tested yet. Another potential source of selection bias might be caused by the voluntary participation in research, but this source of bias is hard to avoid as research participants should always participate on a voluntary basis (58). Individuals who accepted to participate in this telerehabilitation study were probably more motivated by the intervention than the 61.5% individuals who refused which could affect the compliance to the project compared to in clinical settings.

Another potential source of bias is information bias, which consist of bias related to data collection. Different questionnaires and methods of assessments were used in this telerehabilitation project to evaluate pain, function and level of physical activity. As presented in chapter 3, we considered the psychometric properties of the tools selected as well as translated to both English and French. Only the Pediatric Outcomes Data Collection Instrument (PODCI) was already described in children with amyoplasia and the authors concluded that it was valid to evaluate functional outcomes of children with amyoplasia and to compare changes over time (59). Yet, the PODCI was not validated in other types of AMC such as distal arthrogryposis. The sensitivity to detect changes over time and to detect a difference between typically developing children and children with amyoplasia was analysed. The authors analysed changes over a three-year interval and included some participants who underwent different types of surgery which might facilitate the detection of changes. The Physical Activity Questionnaire for Adolescents (PAQ-A) and the Adolescents Pediatric Pain Tool (APPT) were validated for children and adolescents, but not particularly for a population with AMC or for our participants who were older than 18 years old (60, 61). The APPT has been used with children with osteogenesis imperfecta (62) and the PAQ-A with children with cerebral palsy (63). The GAS is a standardized outcome measure used to calculate the extent to which a patient's goals are met. The GAS was developed for a general population to evaluate the results of mental health treatment and community interventions (64). The GAS is an individualized measure who has shown high sensitivity and specificity over rehabilitation intervention (65, 66). The questionnaires selected in this project might have led to a certain degree of information bias as most were not specifically validated for the target population. Nonetheless, the information garnered from this feasibility study serves to guide and inform clinicians and researchers as to the choice of questionnaires, assessment and treatment methods for future studies and perhaps even in the clinical setting. Another potential source of information bias may have been introduced by the virtual measurement of ROM. However, an ICC evaluation was done to mitigate this bias. In order to minimize information bias, evaluators who were not involved with the study intervention performed the measurements. Also, as the data collected was either patient-reported or measured by an evaluator who did not provide the intervention, this source of bias was minimized.

Future directions

This project was the first step in order to assess the effectiveness of an exercise intervention delivered by telerehabilitation with young people with musculoskeletal disorders, more particularly with AMC. The intervention was shown to be feasible and demonstrate promising results for the effectiveness. The following directions are envisaged in future studies: 1) to develop larger studies, 2) Use different approaches of intervention and supervision, and 3) use different teleassessment methods.

Studies with rare disorders are often performed with small sample sizes which make hard to measure the effect of an intervention and to generalize the results to the target population. Given the rarity of AMC, the use of telerehabilitation may facilitate the recruitment of more individuals with rare disorders. This approach may also help to develop multicenter studies with other specialized health care centers, allowing for a larger sample size and appropriate study power. With a larger sample size, higher quality studies can facilitate the development of guidelines and recommendations based on scientific evidence for populations currently underrepresented in the literature.

Telerehabilitation gives the opportunity to develop new types of intervention as well as to study interventions that already exist, but that require frequent follow-ups. New types of intervention involving an interdisciplinary approach or comparing different frequencies of supervision should be studied to determine the benefits. Exercise intervention has been shown to be cost-effective with cerebral palsy and colon cancer compared to usual care, but not for breast cancer (67, 68). Therefore, as benefits and cost may vary depending on the population and based on the type of intervention, the clinicians involved or the frequency of the supervision, a cost-effectiveness analysis should be performed at same time.

Teleassessment is not yet largely studied for musculoskeletal rehabilitation purpose. Objective methods to assess muscle strength, balance or mobility should be developed for when in-person assessment is not possible. As mentioned previously, the use of wearable sensors could be useful, but the feasibility of using those in telerehabilitation intervention needs to be studied. Also, the placement of those sensors needs to be studied before using them with individuals with AMC. Their joint contractures may interfere with accelerometer data due to movement restriction (69). Therefore, the location of those sensors should be adapted for each individual due to their joint contractures.

Chapter 6: Conclusion

Research with individuals with rare disorders remains challenging, but the findings of this thesis show that telerehabilitation is feasible and is a promising approach for youths with rare disorders such as AMC. The goal of developing telerehabilitation intervention is not to replace in-person intervention, but rather to propose an alternative approach for those for who it is difficult to travel to a specialized health care center. As found in the scoping review presented in this thesis, little is known about the effectiveness of an exercise program and this pilot study was the first step in order to evaluate the potential effectiveness of this type of intervention. Results from this pilot study showed promising benefits in regards to an exercise intervention that should be assessed with a randomized control study. Effectiveness of exercise intervention remains to be studied to better evaluate the benefits of this type of intervention on the short term, but also for long-term effects as people with AMC may need to maintain their ROM to prevent functional losses over time (70). A physical activity intervention was not studied in this project as exercise programs were most related to rehabilitation. However, most adults with AMC have a sedentary lifestyle (71) and an early physical activity intervention might help them learning how to move and how they could benefit from an active lifestyle, thus preventing secondary diseases related to a sedentary lifestyle.

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Supplementary materials

These supplementary materials were submitted along with the manuscript presented in chapter 4.

Table S1: Missing value for range of motion measurements

	Not assessed	Assessed in the wrong plane	Not assessed with the same method	Total
Shoulder abduction	2	0	0	2
Shoulder adduction	28	0	0	28
Shoulder flexion	0	12	0	12
Shoulder extension	17	0	0	17
Elbow flexion	0	0	0	0
Elbow extension	0	0	0	0
Forearm pronation	4	0	0	4
Forearm supination	2	0	0	2
Wrist flexion	0	0	2	2
Wrist extension	0	0	2	2
Hip flexion	0	0	2	2
Hip extension	6	1	0	7
Hip internal rotation	9	2	0	11
Hip external rotation	8	2	0	10
Knee flexion	0	0	2	2
Knee extension	0	0	0	0
Ankle dorsiflexion	4	0	0	4
Ankle plantarflexion	4	0	0	4
Total	84	17	8	109

Document S1: Examples of exercise programs

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Programme d'exercices #1

Goals:
1. Self-feed with less compensatory techniques
2. Self-propelling wheelchair
3. Ability to transfer in a car

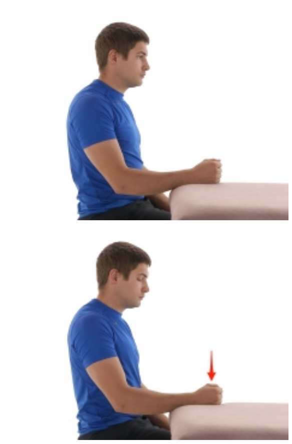
Notes : Repos 1-2 minutes entre chaque série.

1 Flexion coude isométrique

Séries: 2-3 Réps: 10 Tenir: 5 secondes

Placez la main sur le poignet du côté atteint.
Tentez de plier le coude tout en empêchant le mouvement avec votre main.
Il ne devrait pas avoir plus de mouvement.
Assurez-vous de forcer au maximum.

2 Extension iso. du coude



Séries: 2-3 Réps: 5 secondes

Assoyez-vous bien droit devant une table.
Formez un poing avec votre main et placez le poing sur la table, le coude fléchi à environ 90 degrés.
Essayez de déplier le coude en poussant vers le bas contre la table.
Maintenez la contraction pour la durée recommandée, relâchez et répétez.

3 Flexion de hanche, assis

Séries: 3 Réps: 10 Poids: avec assistance de tes propres main; la descente est la phase la plus importante Tempo: 2:3

Assoyez-vous sur une chaise droite avec le dos en position neutre (légèrement arqué).
En gardant le genou fléchi, levez le genou vers la poitrine en ne roulant pas le bassin vers l'arrière quand vous effectuez le mouvement.
Abaissez et répétez.

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4 Flex./ext. coude assis


Séries: 3 Réps: 10 - cette exercice est soit fait avec maman ou avec serviette pour diminuer la friction; idéalement, l'épaule est au meme niveau que le coude

Assoyez-vous.
Appuyez les bras sur la table, les coudes pliés et placez une serviette sous vos mains.
Allongez le bras en glissant la main sur la table.
Poursuivez le mouvement en poussant les épaules vers l'avant.
Revenez à la position de départ en pliant les coudes.

---

5 Extension coude serviette


Séries: 3 Réps: 10 Poids: avec assistance de tes propres main; la descente est la phase la plus importante Tempo: 2:3

Assoyez-vous.
Appuyez un avant-bras sur la table et placez une serviette sous votre main.
Gardez le coude plié.
Allongez le bras en glissant la main sur la table.
Poursuivez le mouvement jusqu'à ce que le coude soit allongé complètement.
Revenez à la position de départ en pliant le coude.

Attention à ne pas forcer avec votre tronc et votre nuque.

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6 Extension hanche sur chaise

Séries: 3 Réps: 10 Poids: avec assistance de tes propres main; la descente est la phase la plus importante Tempo: 2:3

Couchez-vous sur le dos et placez vos jambes sur la chaise.

Décollez le bassin du sol pour faire le pont en serrant les fesses.

Descendez sur le sol et répétez.

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Home Exercise Program #1

Goals:
1. Ability to get in a car
2. Control stair decent
3. Kick higher in karate

Notes : - All exercises should be done on both legs.

1 Step-down

Sets: 3 Reps: 10 Freq: daily Tempo: 2 seconds up, 2 seconds down, SLOW Rest: 1 minute rest in between sets

Stand up straight on a stool supported on one leg. Make sure you have a counter top or railing that is safe and secure beside you for support. You will need to hold throughout.
Align your knee over your toes and slowly beginning to bend at the hips (sending the bum back) and then bending the knee as if you were to sit in a chair.
As you bend, the unsupported leg will lower slowly.
Do not pass your standing knee over your toes and make sure that your heel stays as flat as possible (on the stool).
You do not need to touch the ground; it is more of a mini single-leg squat.
Return to the starting position by unbending the knee of the leg on the stool.
Straighten the knee completely and stand upright before placing the free foot onto the stool.
Repeat.

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2 Hip+knee auto-assisted flexion

Sets: 3 Reps: 10 Freq: daily Rest: 1-2 minutes in between sets

Start in a seated position and surround one knee with your hands.
With the help of your hands, ACTIVELY bring your knee towards your chest and help yourself for the range that is difficult to accomplish.
As you lower your leg, ACTIVELY try to slow the descent as if you wanted to hold your leg in position while gradually releasing your hold.
Repeat.

You may also use a towel or belt looped under your thigh to assist.

3 Standing hip flexion w/ stool

Sets: 2-4 Reps: 15-20 Freq: daily Rest: 1 minute in between sets

Stand up straight beside a chair with a stool in front of you.
Slowly lift your foot up onto the stool, step or textbook by bending your hip, knee, and ankle as much as you can.
Lower your foot to the ground by lifting up and off with control.
When you lift your leg, do not move the rest of your body and attempt not to knock the bench. Your abdominals should be engaged and you should be looking tall and far in front of you.
You can use the number of times you knock the object as a test to track your progress.

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Goal:
To improve walking endurance

Notes:

1- Wall squat - Sets:2-3 / Repetition:5 / Frequency:3x per week at minimum / Hold:10 seconds / Rest:1.5 minute in between sets

Stand up against a wall with your feet in front of you; when your thighs are parallel to the ground your knees should be at appropriately 90 degrees.
Slide down the wall, keeping the upper body in contact with the wall.
Keep the knees in line with the feet. Your knees should not pass your toes and your heels should remain flat on the ground. Do not use your hands.
Slowly stand back up squeezing the buttocks and thigh muscles as you return to your starting position.
Repeat.

2- Bridging - Sets:3 / Repetition:10 / Frequency:3x per week at minimum / Hold:2 second hold while bum is in the air / tempo:2 seconds up / Rest:1 minute in between sets

Lie on your back with your knees bent and one leg crossed over the other.
Contract your abdominal muscles and your buttocks while lifting your buttocks off the ground.
Slowly return to the initial position and repeat.

Progressions possible (in appropriate order):
- place a small stool under supporting limb
- progress to perform with the other leg unsupported (that is actively held, fully extended, in the air)

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3- Clamshell - Sets:3 / Repetition:10 / Weight:use appropriate elastic so that you may complete full movement but last two or three repetitions of each set are difficult / Frequency:3x per week at minimum / tempo:2 seconds open / Rest:1 minute in between each set

Lie on your side with your knees bent and head supported with a resistance band about 1 inch above your knees.
Lift your upper knee without moving your pelvis or lifting the foot off your other foot, in order to create a gap between your thighs.
Lower your knee and repeat.

Once completed, perform the same exercise with the opposite leg on top.

4- Isometric plantar flexion with band and towel (please ignore equipment in image; movement remains the same) - Sets:3 / Repetition:12 / Frequency:3x per week at minimum / Hold:5 second hold / Rest:1 minute rest in between sets

Two exercises to complete, one foot at a time:
1. Against tough elastic band with knee bent to 90 deg

2. Against towel with back supported against a wall and knee fully extended
- make sure that with this exercise is completed without allowing any motion in the ankle (you must match your resistance)

Both exercises involving initiating the movement of pointing your toes.

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Goals:
1. Improve walking endurance
2. Quadriceps strengthening/ reduce pain on thigh

Notes: Walking: Must walk 5 minutes straight and repeat this activity 3 times in one evening. TO BE DONE EVERY DAY.

All other exercises should be done 3x per week.

Recommendations include:
- to walk with braces in home as much as possible and use wheelchair as little as possible
- to walk in the community (ex. push wheelchair in grocery store, at the mall, in school corridors, etc.)
- wear watch at all times, if not, at minimum, when performing home exercise program

1- Bridge - Sets:2 / Repetition:10 / Weight:pillow betweens legs / Hold:5 second hold / tempo:2 seconds to lift bum / Rest:1.5 minutes in between each set

Lie down on your back with your knees bent and a pillow between them.
Lift your bum off the ground. As you lift, squeeze the pillow between your legs.
Hold for a few seconds on top then slowly lower your bum.
Repeat.

Make sure you breathe out every time you lift your bum up.

2- Leg lift - Sets:2 / Repetition:10 / Hold:2 seconds with the leg in the air / tempo:slow on the way up and down / Rest:1.5 minutes in between sets

Lay on your side with your head supported and lower leg bent. Make sure the other hand is holding the ground for stability.
Lift the top leg up as the picture shows, keeping the toes pointing forward and the leg aligned with the body. Make sure you are not rolling forward or backward.
Lower the top leg and repeat.

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3- Sit to stand using STOOL - Sets:2 / Repetition:8 / Rest:2 minutes of rest in between sets

Sit on the stool (placed against a wall to prevent it from moving).
Fold your arms across your chest or simply avoid using your hands.
With your feet slightly apart, lean forward and stand up.
Do not bend your back or jump up.
Slowly sit back down.
Repeat.

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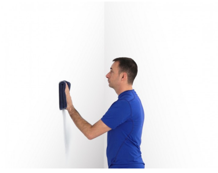
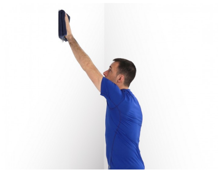
Goals:

1. To use more the left arm during sledge hockey
2. Buttoning up shirt

Notes: To be done: 3x per week at minimum

1- Wall slide + lean into it - Sets:2 / Repetition:10 / Hold:5 seconds





Stand up facing to a wall and place your hand on the wall, with a towel between the wall and your hand.
Slide up as far as you can.
Let yourself lean in towards the wall to feel a stretch under the arm pit area.
Slide your hand back down.
Repeat the exercise.

2- Shoulder flexion with elastic - Sets:3 / Repetition:10 / Weight:red elastic / tempo:4-5 seconds up / Rest:1 mintute in between sets

Tie an elastic band at hip height to a secure object behind you (door knob of closed door) or chair with someone sitting on it.
Start by holding the elastic band with your arm by your side. Do not follow the video for hand placement; it is preferred that you keep a thumbs up position by making a fist around the elastic (do not loop it around your hand or wrist).
Focus on keeping your arm locked the entire time (do not let your wrist fall or your elbow bend).
Stand as far forward as you can so that you may feel a small stretch in the front of your shoulder.
Raise your arm in front of you to nose or eye level.
Slowly return to the starting position.

3- Assisted shoulder extension with hockey stick - Sets:2 / Repetition:10 / Hold:5 seconds

Stand straight with your chin tucked in holding a stick.
Place your left arm along your side and the other arm straight in front.
Push on the stick with your right arm to push the left shoulder back as high as you can, without poking your neck forward or causing pain.
Maintain the position, lower your arm and repeat.

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4- Grip strength with ball - Sets:3 / Repetition:10 / Hold:3 seconds / Rest:1 minute in between sets

Remember to have your forearm supported by the table in front of you.
The side of your hand with your little finger should be resting on the table.
Hold a small ball in your hand and squeeze it as hard as you can.
Hold 3 seconds.
Release the ball slowly.
Repeat.

5- Full grip with putty - Weight:once putty received; fluorescent red

Make a fist with your fingers and thumb as you squeeze the putty. Make sure your forearm is supported on the table and that you are keeping your wrist as close to a 'thumbs up' position as possible.
Dig all the fingers in.
Hold 3 seconds.
Release grip.
Reshape the putty, if necessary, and repeat.

6- Standing plantar flexion - Sets:3 / Repetition:15 / tempo:2 seconds up

Stand on both feet on the floor next to a chair or table (for balance purposes).
Come onto the toes (raise heels) of both feet without bending the knee.
Keep the bum, tummy and thighs tucked tight.
Slowly bring your heels back down and repeat.

Rest between 1-2 minutes in between each set.

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Notes: Recommendations:

- be conscious of good sitting posture (steps to take: tuck in tummy by drawing belly button in without cutting off your breath, bum shift to left and align shoulders with hips)
- continue regular physiotherapy exercises (emphasis on hip flexor stretching and adductor range of motion)
- remember to exhale during strenuous part of each exercise

1- Seated hip flexion - Sets:2 / Repetition:10 each leg / Frequency:2-3x per wk / tempo:2 seconds up / Rest:2 minute rest in between sets

Sit on a straight chair with your back in neutral position.
Keeping your knee bent, lift your thigh off the chair as high as you can. Do not roll your pelvis backwards or lean back into your chair as you lift.
Lower and repeat.

Assistance required: helper
1. minimal on L
2. moderate on R

2- Hip adductor strengthening - Sets:3 / Repetition:5 / Frequency:2-3x per week / Hold:5 seconds / Rest:2 minute rest in between sets

*** PLEASE IGNORE VIDEO. FOLLOW INSTRUCTIONS AND REFER TO EXERCISE POSITION.

Lie onto your back.
Have someone place your knees and hips in a bent position so that your feet are practically flat on the floor. (crook-lying position)
Engage your core by contracting your abdominal muscles.
Keep your legs in this position, with your knees pointing up at the ceiling, squeezing the inside of your thighs together x 5 seconds.
Slowly let your legs fall into the hands of your helper.
Repeat.

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3- Clamshell - Sets:2 / Repetition:10 each side / Frequency:2-3x per wk / tempo:2 seconds up / Rest:2 minute rest in between sets

Lie on your side with your knees bent and head supported.
Elevate the top leg slightly so it doesn't touch the bottom leg.
Keep your top foot directly above the bottom one and lift/rotate your top knee without rolling back.
Lower your knee and repeat.
Breathe normally during this exercise.

4- Hip extension strengthening - Sets:3 / Repetition:5 each leg / Frequency:2-3x per wk / tempo:2 seconds up / Rest:2 minutes in between sets

Lay with the face down and the affected side knee bent at 90 °. Your helper should help ensure the best alignment possible, meaning thigh brought in and foot up towards the ceiling as displayed. The best way to accomplish this is by placing one hand under the front of thigh and the other hand holding the lower leg, just above the ankle .
Lift the thigh off the surface.
Slowly lower to the start position.
Repeat the exercise.

5- Passive hip internal rotation stretch - Sets:3 / Frequency:2-3x per wk / Hold:30 seconds / Rest:30 seconds-1 minute in between

Place one hand under the knee and cup the heel with your other hand.
Using both hands, bend the hip and knee up until the thigh is perpendicular to the body and the shin is perpendicular to the thigh, all the while keeping the knee in midline.
Move one hand to the outside of the knee and the other to the inside of the heel and rotate the lower leg as far as possible towards the OUTSIDE without letting the knee fall in towards the patient's midline.
Hold x 30 seconds.
Come back to center and release.

* NO NEED TO PERFORM HIP EXTERNAL ROTATION STRETCH IN OTHER DIRECTION (foot towards inside) AS DEPICTED IN THE VIDEO.

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Objectives:

1. Manual dexterity
2. Hand endurance
3. Standing endurance

Notes: Recommendations:

- si l'élastique ou pâte à modeler utilisé devient facile, vous pouvez progresser la résistance d'un niveau (putty rose fluo à vert, élastique facile à moyen à difficile)
- à faire programme d'exercice 2-3x par semaine pour débuter

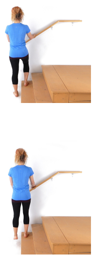
N'hésitez pas si vous avez des questions!

1- Montée/descente de marche - Séries:2-3 / Répétition:10 / tempo:2 sec. montée / Repos:1 minute entre séries du même côté

Tenez-vous sur une marche avec la jambe à renforcer.
Placez la majorité du poids du corps sur le talon de la jambe d'appui.
Inclinez votre poids sur la jambe sur la marche.
Montez sur la marche lentement, tout en gardant le genou aligné vers l'avant (non l'extérieur ou l'intérieur) et les hanches au niveau. Gardez le dos droit; évitez de pencher le tronc.
Descendez en ramenant la jambe libre vers le sol.

Répétez avec la jambe opposé. Suggérons de tenir avec minimum une main pour débuter et favoriser un bon contrôle du mouvement.

2- Montée de marche latérale - Séries:2-3 / Répétition:10 / Tenir:2 sec. montée / Repos:1 min. entre séries du meême côté

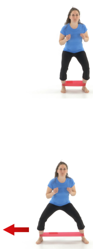


Tenez-vous de côté à un escalier. Suggérons de tenir avec une main au minimum pour favoriser un bon contrôle excentrique (phase descendante).
Montez la marche avec la jambe du coté de la marche, en gardant le genou aligné vers l'avant et les hanches au niveau. Utilisez le talon de la jambe d'appui pour monter plutôt que de pousser avec la jambe libre.
Descendez en ramenant la jambe saine vers le sol.

JAMBE DROITE:
- utilisez livres solides, placé de manière sécuritaire, proche d'une rampe ou un meuble ou chaise solide

JAMBE GAUCHE:
- utilisez l'escalier standard

3- Pas latéraux avec élastique - Séries:4 / Répétition:10 pas allez / Poids:élastique facile / tempo:2 secondes par pas / Repos:1-2 minutes



Placement de l'élastique au-dessus des genoux devrait être fait avec pieds collés.
Débutez en position de squat partielle avec un élastique autour des CUISSES (ne vous fier pas à l'image).
Effectuez des pas latéraux.
Gardez les genoux pointant vers l'avant et pieds écartés la largeur du bassin.
Chaque pas doit couvrir environ 50% de la distance initiale entre les deux pieds.

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4- Opposition avec pâte - Séries:3 / Répétition:5

Opposez le pouce avec chaque doigt séparément et serrez la pâte entre les deux.
Répétez ou maintenez selon les directives.

À faire:
1. pouce + index
2. pouce + majeure
3. pouce + annulaire
4. pouce + auriculaire

5- Crochet des doigts - Séries:3 / Répétition:5

Faites un crochet avec les doigts en serrant la pâte.
Utilisez seulement les quatre doigts.
Répétez ou maintenez selon les directives.

6- Serrer le poing, pâte - Séries:3 / Répétition:10

Serrez la pâte avec les doigts et le pouce.
Répétez ou maintenez selon les directives.

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7- Adduction pouce, pâte - Séries:3 / Répétition:10


Placez la pâte dans la paume et fléchissez les doigts légèrement. Avec le pouce, poussez dans la pâte contre l'index.
Ne poussez pas sous l'index.
Remodelez la pâte et répétez.

8- Adduction doigts, pâte - Séries:2 / Répétition:5


Serrez vos doigts ensemble sans les plier contre la pâte à modeler.

À faire:
1. index + majeure
2. majeure + annulaire
3. annulaire + auriculaire

9- Abduction des doigts avec pâte - Séries:3 / Répétition:8


Placez la pâte autour d'une paire de doigt à la fois puis tentez de les écarter.
Remodelez la pâte et répétez.

À faire:
1. index + majeure
2. majeure + annulaire
3. pouce + index

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Document S2: Infographic of the telerehabilitation project

Using technology to remotely deliver a home exercise program to youths with AMC

What is this study about ?
To evaluate the feasibility and to explore the effectiveness of a Home Exercise Program (HEP) for youths with AMC using telerehabilitation

What is AMC ?
An umbrella term describing over 400 conditions characterized by congenital joint contractures present in at least two body areas, occuring in 1 of every 4300 to 5100 live births

What is telerehabilitation ?
An innovative method to deliver rehabilitation services remotely

VIDEO CONFERENCE
PHONE CALLS

Decreases hospital visits

DECREASE COST
EASIER ACCESS

Why use it ?
To deliver regular interventions with frequent follow-ups, such as exercise programs

TRANSPORTATION FREE
SAVES TIME

To provide sub-specialized healthcare to individuals who live far

Recruitment
Invitations to participate were done in clinic, and using phone calls, postal invitations, and through the Canadian AMC support group on social media

To identify potential participants, medical records were reviewed

Participants were recruited from different provinces across the country

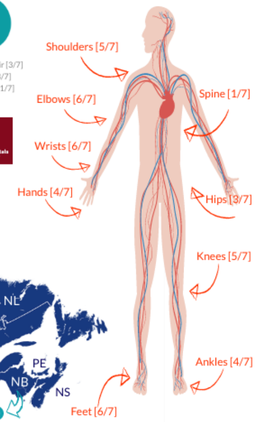
Participants
11 youth initially consented
7 completed the program

Median age 16.9 years

Mobility

Wheelchair [3/7]
Walking [3/7]
Crutches [1/7]

AMC Joint Involvement



Data Collected
Telerehabilitation meetings were provided remotely using Zoom pro:

- Assessments
- Program delivery
- Follow ups

EVERY 3 WEEKS

12 WEEKS

Participants were asked to perform the HEP for 15 - 20 mins each session

2 to 3 x WEEK

The HEP was developed using personalized goals that were set with each participant

DRESSING
EATING
WALKING UP STAIRS

TO INCREASE ENDURANCE WITH

WALKING
STANDING
MANUAL DEXTERITY/ WRITING

TO BE MORE INDEPENDENT WITH

Pain was measured for all 7 participants:

- 4 reported pain before the HEP

TO MANAGE PAIN

Results

Feasibility
Technical issues were:

- Present in 3 out of 35 meetings
- All resolved during the same meeting

ECHO AND SOUND DIFFICULTIES
LOST OR WEAK CONNECTION

Satisfaction
All participants were either satisfied or very satisfied with:

- The remote evaluation
- Delivery of the HEP
- Follow ups and overall organization

7/11
63.6%
COMPLETION

Home Exercise Program
69.4%
COMPLIANCE

Effectiveness
The following improvements were noted at the end of the 12 week HEP:

Pain Scale Used for APPT
A LOT OF PAIN
NO PAIN

APPT – Adolescent Paediatric Pain tool
Clinically significant reduction of pain (-2 points)

PODCI – Paediatric Outcomes Data Collection Instrument
Improvements in pain and comfort levels

PAQ – Physical Activity Questionnaire
Overall improvements in physical activity levels

GAS – Goal Attainment Scale
Each participant had 1 - 3 goals for a total of 15 goals

12/15 GOALS ATTAINED

What's next?
Integration of telerehabilitation in clinic to facilitate the delivery of personalized interventions, such as a HEP

Interdisciplinary approach to help youth achieve their goals in a comprehensive manner

Why is this important?
This pilot study established the feasibility of using telerehabilitation for remote delivery of an individualized exercised program

The overall results of the HEP showed that most of the youth attained their goals

Podcast
Click to tune in for testimonials from our community

Poster
Click to view a scientific summary of our study

References
Gagnon M, Collins J, Elfassy C, Marino Merlo G, Marsh J, Sawatzky B, Yap R, Hamdy R, Veilleux LN, Dahan-Oliel N. (2020). A Telerehabilitation Intervention for Youths With Arthrogryposis Multiplex Congenita: Protocol for a Pilot Study. JMIR Res Protoc, 9(6):e18688. doi: 10.2196/18688.

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