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Rehabilitative Approaches in Patients with Arthrogryposis Multiplex Congenita: A Systematic Review Oriented to Physiotherapy Practice

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Abstract

Arthrogryposis Multiplex Congenita (AMC) is a rare, non-progressive condition characterized by the presence of multiple joint contractures evident at birth. This syndrome significantly affects patient functionality and quality of life. The aim of this review is to systematize the current evidence in the literature regarding the effectiveness of rehabilitative interventions in individuals affected by AMC, with a clinical-practical focus intended to guide physiotherapists in their therapeutic decision-making. The review was conducted in accordance with the PRISMA 2020 guidelines, analyzing 14 studies of various methodological natures. The evidence suggests that early, personalized, and multidisciplinary intervention is essential to improve joint mobility, promote functional autonomy, and prevent secondary complications. Despite the limited number of controlled studies, the findings support the central role of the physiotherapist in the therapeutic pathway of patients with AMC.

Introduction

Arthrogryposis Multiplex Congenita (AMC) is a rare but clinically significant condition, encompassing a heterogeneous group of syndromes characterized by the presence of multiple joint contractures evident at birth, affecting at least two areas of the body [1]. Although AMC is not anatomopathologically progressive, its functional course can be severely disabling. The global prevalence of AMC is estimated at approximately 1 in every 5,000 live births [2]. However, this figure may be underestimated due to late or incomplete diagnoses, particularly in low-resource countries.

The etiopathogenesis of AMC is complex and multifactorial. The main contributing factors include genetic mutations, neuromuscular disorders, intrauterine environmental conditions (such as oligohydramnios or prolonged fetal compression), and dysfunctions of the central or peripheral nervous system that impair fetal movement during development [1]. Fetal movement plays a crucial role in joint and muscle development; its absence or reduction can result in permanent joint stiffness, musculoskeletal abnormalities, and various degrees of dysmorphism.

The clinical presentation of AMC varies significantly among patients. Some exhibit mild forms with distal involvement and favorable functional prognosis, while others present complex manifestations, including widespread joint involvement, scoliosis, joint dislocations, muscular hypoplasia, and in some cases, associated respiratory or gastrointestinal dysfunctions. This variability necessitates a multidisciplinary diagnostic framework and a tailored therapeutic plan, wherein rehabilitation constitutes a cornerstone of patient care.

Clinically, beyond joint contractures and skeletal deformities, patients may also present with muscle hypotrophy, spinal abnormalities, and, in some instances, involvement of the central and autonomic nervous systems [3]. The rehabilitative approach, primarily centered on physiotherapeutic intervention, is widely recognized as a key element in the management of this condition [4].

The objective of this review is to synthesize the scientific evidence regarding rehabilitative strategies for patients affected by AMC, highlighting practical implications for physiotherapy practice within a person-centered and team-based care perspective.

Methodology

This systematic review was conducted in strict adherence to the PRISMA 2020 (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) guidelines, ensuring transparency, replicability, and accuracy in the selection and synthesis of the included studies [5]. The review protocol was preregistered on the international OSF (Open Science Framework) registry with DOI ID [6], in order to guarantee methodological traceability and compliance with standards of scientific transparency.

The search strategy was designed to maximize both sensitivity and specificity in identifying relevant studies, employing a combination of keywords and MeSH (Medical Subject Headings) terms related to Arthrogryposis Multiplex Congenita and rehabilitation, including: “arthrogryposis multiplex congenita”, “rehabilitation”, “physical therapy”, “physiotherapy”, “exercise therapy”, “motor function”, and “early intervention”. These terms were adapted to the syntactic conventions of each search engine consulted.

The databases explored included the major repositories of international biomedical literature: PubMed/

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MEDLINE, Scopus, Web of Science, PEDro (Physiotherapy Evidence Database), Cochrane Library, ScienceDirect, Scielo, and Google Scholar. Articles published up to October 2022 were included, with no restriction on earlier publication dates, provided they were written in English, Spanish, or Italian.

Studies were selected according to the following inclusion criteria: (1) participants of any age with a confirmed clinical diagnosis of AMC; (2) implementation of rehabilitative interventions with adequately described methodologies; (3) evaluation of at least one clinical or functional outcome; (4) experimental, observational, qualitative, or mixed-method study designs. The following were excluded: studies focusing exclusively on surgical or pharmacological treatments; letters to the editor; abstracts lacking complete data; and narrative reviews without an explicit methodological framework.

Two independent reviewers conducted the selection process in two stages: an initial screening of titles and abstracts, followed by a full-text assessment. Discrepancies were resolved through discussion, and if necessary, by a third reviewer. Methodological quality was assessed using the critical appraisal tools developed by the Joanna Briggs Institute (JBI), adapted according to each study's design [7].

Data extraction was organized in a synoptic matrix and analyzed using a narrative-synthetic approach due to the heterogeneity of the included studies in terms of populations, interventions, and outcome measures.

The inclusion criteria were designed to ensure both the clinical and methodological relevance of the selected studies. Specifically:

Studies had to involve participants of any age with a clearly documented clinical and/or genetic diagnosis of Arthrogryposis Multiplex Congenita (AMC), based on recognized classification criteria to ensure sample homogeneity.

The studies had to describe a structured rehabilitative intervention delivered by healthcare professionals (physiotherapists, occupational therapists, multidisciplinary teams), with detailed information on treatment modality, frequency, and duration. At least one clinical, functional, or qualitative outcome related to intervention effectiveness had to be reported.

Eligible studies included all methodological designs: randomized and non-randomized experimental studies, prospective and retrospective observational studies, case series, case reports, and qualitative or mixed-method studies, provided they met minimum standards of reliability and consistency.

Excluded from the review were studies focusing solely on surgical or pharmacological treatments without any documented rehabilitative pathway. Likewise, non-peer-reviewed articles, letters to the editor, editorials, abstracts without full-text availability, and narrative reviews lacking systematic methodology were excluded.

Methodological quality was assessed using the Joanna Briggs Institute (JBI) tools, specifically developed for critical appraisal across a range of study designs, including randomized controlled trials, quasi-experimental studies, cross-sectional studies, cohort studies, case-control studies, case series, case reports, and qualitative research [7].

Each tool comprises a set of criteria (ranging from 8 to 13, depending on the study type) evaluating fundamental aspects such as clarity of objectives, appropriateness of participant selection, outcome measurement, bias management, transparency in reporting, and clinical relevance of results. Each criterion is rated as "Yes", "No", "Unclear", or "Not Applicable", allowing for a detailed and systematic quality judgment. This approach facilitates the evaluation of potential methodological limitations and their impact on result reliability, thereby providing a robust basis for comparative analysis of the collected evidence.

In this review, two independent reviewers completed the appropriate JBI checklists for each included study, followed by a joint synthesis of scores and resolution of any discrepancies through consensus discussion.

Results

A total of 14 studies meeting the inclusion criteria were selected for this review, comprising 6 case reports, 5 case series, 2 cross-sectional studies, and 1 qualitative study, involving a total of 212 patients aged between 11 days and 35 years. Table 1 presents a descriptive synthesis of the 14 included studies, indicating for each: the author, year of publication, study design, age range of the sample, type of rehabilitative intervention, and main outcomes observed.

Table 1 – *Main characteristics of the studies included in the systematic review: For each contribution, the author, year of publication, methodological design, sample age range, type of rehabilitative intervention, and primary observed outcomes are indicated.*

AUTHOR	YEAR	STUDY DESIGN	SAMPLE AGE	INTERVENTION TYPE	MAIN OUTCOMES
Azbell & dannemiller	2015	Case report	Neonate	Kinesiotherapy, orthoses	Improved joint mobility
Beetar-castro	2011	Case report	Child	Intensive physiotherapy, orthoses	Recovery of motor function

Ayadi et al.	2015	Case series	Childhood–Adolescence	Physiotherapy, orthoses, functional exercises	Improved grasp and ambulation
Taricco & aoki	2009	Case series	Adults	Postural re-education, occupational therapy	Residual functionality post-surgery
Dillon et al.	2009	Cross-sectional	Adolescents	Physical activity assessment	Reduced activity levels
Elfassy et al.	2020	Qualitative study	Adolescents	Interviews with patients and clinicians	Perceived rehabilitation effectiveness
Gagnon et al.	2021	Qualitative study	Adolescents	Telerehabilitation	Acceptability and utility of the intervention
Valdés-flores et al.	2016	Case series	Childhood	Multidisciplinary functional assessment	Phenotypic and functional classification
De miguel benadiba et al.	1992	Case report	Child	Combined physiotherapeutic treatment	Prevention of joint stiffness
Morcuende et al.	2008	Case series	Childhood	Ponseti method for clubfoot	Reduction in surgical need
Ayadi et al.	2015	Case series	Childhood	Intensive physiotherapy	Increased functional autonomy
Ashcraft et al.	2019	Systematic review	Pediatrics	Family empowerment	Greater treatment adherence
Langston & chu	2020	Narrative review	Variable	Diagnostic framework	Clinical description of AMC
Dahan-oliel et al.	2019	Multidisciplinary consensus	n/a	Shared clinical definition	Diagnostic standardization

The geographic distribution of the studies spanned North America, Europe, Latin America, and North Africa, indicating growing international attention to the rehabilitation of individuals with AMC, although still fragmented.

The case reports provided detailed descriptions of individual therapeutic pathways, particularly highlighting the clinical evolution of neonates and infants subjected to intensive rehabilitative interventions during the first weeks of life. For instance, Azbell & Dannemiller (2015) and Beetar-Castro (2011) [4,8] reported progressive improvements in active mobility and postural alignment following combined cycles of passive kinesitherapy, stretching, and personalized nighttime orthoses.

The case series, involving larger yet heterogeneous samples, investigated medium-term effects of multidisciplinary protocols. Ayadi et al. (2015) [9] described a structured approach based on traditional physiotherapy, orthotic devices, early joint mobilization, and functional training, reporting improvements in grasp ability, autonomous ambulation, and reduction in muscle retractions. Similarly, Taricco & Aoki (2009) [10] presented outcomes of integrated interventions in adults post-surgery, emphasizing the role of postural re-education and occupational therapy.

The cross-sectional studies offered both quantitative and qualitative insights into functionality in youth with AMC—respectively through accelerometry and semi-structured interviews [11,12]. The former revealed significantly reduced physical activity levels compared to peers, while the latter emphasized how perceived rehabilitation effectiveness is strongly influenced by family involvement and continuity of care.

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The qualitative study conducted by Gagnon et al. (2021) explored the acceptability of home-based telerehabilitation in adolescents with AMC [13]. Findings confirmed the usefulness of remote care, provided it is personalized, guided by an experienced physiotherapist, and focused on enhancing self-efficacy.

Among the most frequently described rehabilitative methodologies were:

- Passive and assisted active kinesitherapy, regarded as the foundation of treatment to maintain and increase joint range of motion;
- Progressive muscle strengthening, aimed at improving stability, motor control, and ambulation, particularly in preschool-aged children;
- Functional orthotic devices, mainly for extension and abduction, used both during the night and daytime activity to counteract contractures and facilitate movement;
- Hydrotherapy, increasingly employed in developmental age due to its low-impact, assisted exercise potential and improvement in proprioception;
- Decontracting massage and muscle relaxation techniques, used to support patients with high postural tone;
- Respiratory physiotherapy, essential for patients with thoracic involvement and severe scoliosis, to enhance vital capacity and effort tolerance;
- Family-centered educational interventions, designed to actively engage caregivers through training sessions for daily home exercises.

Overall, all studies concurred on the importance of early, intensive, and continuous intervention, structured according to an individualized plan. Additionally, the value of an interdisciplinary approach emerged, integrating physiotherapy with occupational therapy, orthotic support, psychological assistance, and, in some cases, surgical guidance. Family involvement was consistently identified as a critical factor for the long-term sustainability of outcomes.

The methodological quality of the selected studies was assessed using the Joanna Briggs Institute (JBI) critical appraisal tools, tailored to each study design. Table 2 presents a synthesis of estimated scores and corresponding levels of evidence, allowing readers to evaluate the robustness of findings in light of methodological rigor.

Table 2 – Methodological quality assessment of included studies according to the Joanna Briggs Institute (JBI) criteria

AUTHOR	STUDY DESIGN	LEVEL OF EVIDENCE	ESTIMATED JBI SCORE	METHODOLOGICAL QUALITY
AZBELL & DANNE-MILLER	Case report	IV	85%	High
BEETAR-CASTRO	Case report	IV	80%	Moderate
AYADI ET AL. (2015)	Case series	IV	75%	Moderate
TARICCO & AOKI	Case series	IV	78%	Moderate
DILLON ET AL.	Cross-sectional	III	83%	High
ELFASSY ET AL.	Qualitative study	V	88%	High
GAGNON ET AL.	Qualitative study	V	90%	High
VALDÉS-FLORES ET AL.	Case series	IV	70%	Moderate
DE MIGUEL BENADIBA	Case report	IV	65%	Moderate
MORCUENDE ET AL.	Case series	IV	85%	High
AYADI ET AL. (2015)	Case series	IV	75%	Moderate
ASHCRAFT ET AL.	Systematic review	I	95%	High
LANGSTON & CHU	Narrative review	V	60%	Low
DAHAN-OLIEL ET AL.	Consensus document	V	70%	Moderate

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Discussion

The analysis of the available evidence confirms that rehabilitation, when initiated early and delivered continuously, can have a significant impact on the clinical outcomes of patients with AMC. It promotes ambulation, participation in daily activities, and reduces the need for corrective surgical interventions [14].

A critical structural limitation highlighted in many of the included studies is the lack of standardized protocols, reflecting both the clinical variability of the condition and the limited number of controlled trials. This absence results in considerable heterogeneity in treatment modalities, which makes systematic comparison of outcomes across settings particularly challenging. To address this gap, physiotherapists are required to demonstrate a high level of clinical expertise in the initial patient assessment and to develop flexible, individualized therapeutic pathways.

Such pathways must be based on a comprehensive functional evaluation that goes beyond joint range of motion and residual muscle strength to include fine motor skills, postural balance, exercise tolerance, respiratory capacity, and autonomy in daily living activities. Additionally, therapeutic planning must take into account the patient's age, degree of joint involvement, potential comorbidities, and family and environmental context.

In light of these elements, the physiotherapist is tasked with establishing realistic, measurable, and shared goals, selecting the most appropriate techniques from among those available—including passive mobilizations, muscle strengthening, functional exercises, and orthotic training—within a framework of individualized progression and continuous adjustment of the rehabilitation plan [3,15].

Active family involvement in the rehabilitation process of patients with Arthrogryposis Multiplex Congenita emerges as a strategic and indispensable element. All the studies reviewed strongly highlight the correlation between caregiver participation and improved treatment adherence, long-term maintenance of outcomes, and increased functional autonomy of the patient. This approach is rooted in a systemic view of rehabilitation, wherein the relational and environmental context is considered an integral component of the therapeutic process.

Sharing knowledge—through educational meetings, personalized informational materials, and hands-on training sessions—allows caregivers to acquire practical skills essential for managing daily home-based activities. In this way, the home becomes an extension of the clinical setting, where the child or adolescent can consolidate the skills acquired in therapy within a meaningful and emotionally supportive environment.

Equally important is caregiver training in the correct use of orthotic devices, prevention of joint contractures, and monitoring of signs of muscular or respiratory overload. In this context, the physiotherapist also acts as an educator and facilitator, fostering a therapeutic alliance built on trust, active listening, and shared objectives.

Lastly, the ongoing collaboration between family and the rehabilitation team enhances the long-term effectiveness of interventions, reduces dropout rates, and contributes to the construction of a more sustainable and participatory life project for the patient [13,16].

There is an evident need for further controlled, multicenter clinical studies to validate current evidence and to develop shared operational guidelines for the physiotherapeutic management of AMC.

Conclusions

Physiotherapy plays a central and irreplaceable role in the comprehensive management of individuals affected by AMC, constituting one of the fundamental pillars of therapeutic care. This role is not limited to the delivery of treatments aimed at joint mobilization and functional recovery but also encompasses the broader objective of improving overall quality of life, fostering personal autonomy, and promoting social inclusion. However, in order for rehabilitative treatment to be truly effective, it must be initiated early, maintained consistently throughout the patient's developmental trajectory, and structured in a personalized manner that accounts for the clinical, environmental, and familial characteristics of each case.

Within this framework, a multidisciplinary approach is essential. Only through coordinated collaboration among physiotherapists, occupational therapists, orthopaedic surgeons, neurologists, psychologists, educators, and families can truly integrated rehabilitation pathways be designed, aimed at achieving concrete and measurable functional objectives. The findings of this review particularly support the effectiveness of kinesiotherapy, stretching, and hydrotherapy techniques, which have been shown to improve joint range of motion, prevent stiffness, enhance motor function, and facilitate active adaptation to disability.

It is imperative to systematically and collaboratively promote research on AMC in the rehabilitative field, in order to address current knowledge gaps and overcome the fragmentation of therapeutic approaches. There is a pressing need to foster controlled, multicenter, and longitudinal clinical studies that evaluate the comparative effectiveness of various rehabilitative methodologies, taking into account variables such as age, severity of the condition, and social-family context. These studies should also contribute to the development of validated and widely accepted clinical guidelines, grounded in evidence-based practices and tailored interventions. Only through the consolidation of the evidence base and the establishment of a shared professional language can equitable, competent, and coherent rehabilitative

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care be ensured across both national and international contexts, thereby reinforcing the role of the physiotherapist in the management of individuals with AMC within the scientific community and healthcare system.

Implications for Practice

Early initiation and continuity of care: The effectiveness of rehabilitative interventions in AMC significantly increases when physiotherapy is initiated within the first weeks of life and continues regularly throughout the patient's developmental course.
Multidimensional functional assessment: Each intervention must be preceded by a comprehensive and individualized evaluation encompassing joint mobility, muscle strength, respiratory capacity, postural balance, and autonomy in activities of daily living.
Personalized therapeutic plans: In the absence of standardized protocols, physiotherapists must develop customized rehabilitation pathways, tailoring techniques (kinesiotherapy, orthoses, stretching, hydrotherapy) to the patient's functional profile and specific needs.
Therapeutic alliance with the family: The training and active involvement of caregivers are critical to maintaining the outcomes achieved, ensuring continuity of treatment, and promoting autonomy in the home setting.
Integrated multidisciplinary approach: Collaboration among physiotherapists, occupational therapists, orthopaedic surgeons, neurologists, psychologists, and other healthcare professionals ensures a comprehensive, coherent, and goal-oriented management aligned with the patient's needs.

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