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PHYSIOTHERAPY APPROACH IN THE EVALUATION AND TREATMENT OF RARE DISEASES: AN INTEGRATIVE REVIEW

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ABSTRACT

Rare diseases are conditions of low prevalence, that is, they affect up to 65 out of every 100,000 individuals. Many rare diseases are incurable, with repercussions on various body systems, affecting quality of life and functionality due to characteristics such as chronicity, progression and often the lack of specific medications. Rehabilitation is a way to mitigate the symptoms and progression of the disease. Thus, the aim of the study is to analyze the available evidence on the physiotherapeutic approach in the evaluation and treatment of rare diseases, and its impact on the quality of life and functionality of people with rare diseases. This is an integrative review carried out with articles published between the years 2014 -2024 through the electronic databases PEDro, Virtual Health Library, Cochrane and PubMed. Repeated articles that did not use physiotherapy as a therapeutic approach and that were not indexed in free platforms were excluded, totaling an analysis of 14 articles with a sample of 452 individuals, with 20 different pathologies. Six studies addressed quality of life and five assessed muscle strength. Other outcomes observed were depression, aerobic capacity, and body composition. Ten studies analyzed used motor physiotherapy as an approach. Despite the characteristics of rare diseases, physiotherapy, regardless of the modality practiced, is a great ally in the treatment of these individuals. Since it promotes positive effects on your functional capacity and consequently on all aspects of quality of life.

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INTRODUCTION

In Brazil, rare diseases (RDs) are those that affect 65 out of every 100,000 individuals, or less than 1 in every 1,539 live births, and very rare diseases are those that affect 1 in every 1 million live births (Cortés-Martín, Sánchez-Garcia, Rodríguez-Blanque, 2022; Long et al., 2022; Brazil, 2024).

Currently, there are between 6 and 8 thousand pathologies considered rare. They are usually chronic, progressive and degenerative, leading to dysfunction and impairment in quality of life. Of these, 80% are genetic and 20% are related to environmental, infectious, immunological factors, among others. RDs usually do not have a treatment that leads to a cure, leaving those affected with palliative care and rehabilitation services as alternatives to mitigate symptoms (Brasil, 2022).

The diagnosis is usually complex and time-consuming, causing multifaceted impacts on the biopsychosocial sphere of individuals. Physical, mental and behavioral changes resulting from the disease compromise the quality of life of the patient and their families, who depend on several specialized health services and follow-up with a multiprofessional team (Luz; Silva; DeMontigny, 2016). In addition, most RDs lack well-established diagnostic protocols. Many do not have recognized therapy and, in certain cases, pharmacotherapy has high costs, making it unfeasible to acquire it for many families. (Chung et al., 2022).

According to the World Health Organization - WHO (2012), Quality of Life is defined as the individual's perception of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards and concerns, and although many genetic diseases have no treatment, the care offered by a multidisciplinary team offers emotional support and fundamental directions for follow-up, both for caregivers and patients. However, the difficulty in diagnosis, with the need for numerous trips to and from health centers, can result in the rapid advancement of diseases due to lack of treatment, impacting the quality of life of these individuals (Iriart et al., 2019).

According to the International Classification of Functioning, Disability and Health, functionality and disability are related to health conditions, considering not only the functions of the organs or structures of the body, but also the limitations of activity and social participation in the environment where the individual lives and lives (WHO, 2020).

The objectives of a rehabilitation conduct must be established together, and to be concrete and feasible, they must be centered on the functional needs of patients, aiming to

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improve their activity and participation through exercises, adjustments, and adaptations of the environmental context (Silva et al., 2024).

According to O'Sullivan, Schimitz and Fulk (2018), to draw up a rehabilitation plan, the physiotherapist must use clinical reasoning, which encompasses various mental skills to analyze data, make choices and define treatment strategies. Clinical decisions are the result of this process and underpin patient care.

Several elements, such as professional experience, technical knowledge, and patient particularities, play important roles in decision-making. A good therapist can perceive, through evaluation, barriers and facilitators in the different aspects of the user's life. Based on this, measures to reduce or enhance these factors are included in the treatment plan. Follow-up outcomes are analyzed by taking into account the patient's progress toward the goals set and the expected outcomes set out in the treatment plan (O'Sullivan, Schmitz, Fulk, 2018).

Due to the low incidence, it is challenging to bring together a patient population to conduct comprehensive research studies on a given disease. The treatment of these conditions is centered on the maintenance of different clinical complications and the consideration of the individual needs of each patient, aiming to improve their quality of life. To ensure the proper management of rare diseases, it is crucial to keep information up to date, including case registration and description, as well as the development of specific guidelines and protocols to improve clinical practice. A multidisciplinary approach is key in the context of rare diseases and should consider perspectives from all areas of health, including physiotherapy (Cortés-Martín, Sánchez-García, Rodríguez-Blanque, 2022).

Thus, this integrative review aims to analyze the available evidence about the physiotherapeutic approach in the evaluation and treatment of rare diseases, and its impact on the quality of life and functionality of individuals affected by these pathologies.

METHODOLOGY

This is an integrative review, following some of the criteria established by the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA-Checklist).

The guiding question was "what are the effects of physiotherapy, in any modality, on the quality of life and functionality of patients with rare diseases?". Using the PICO strategy, the population (P) corresponds to people with rare diseases; intervention (I) being

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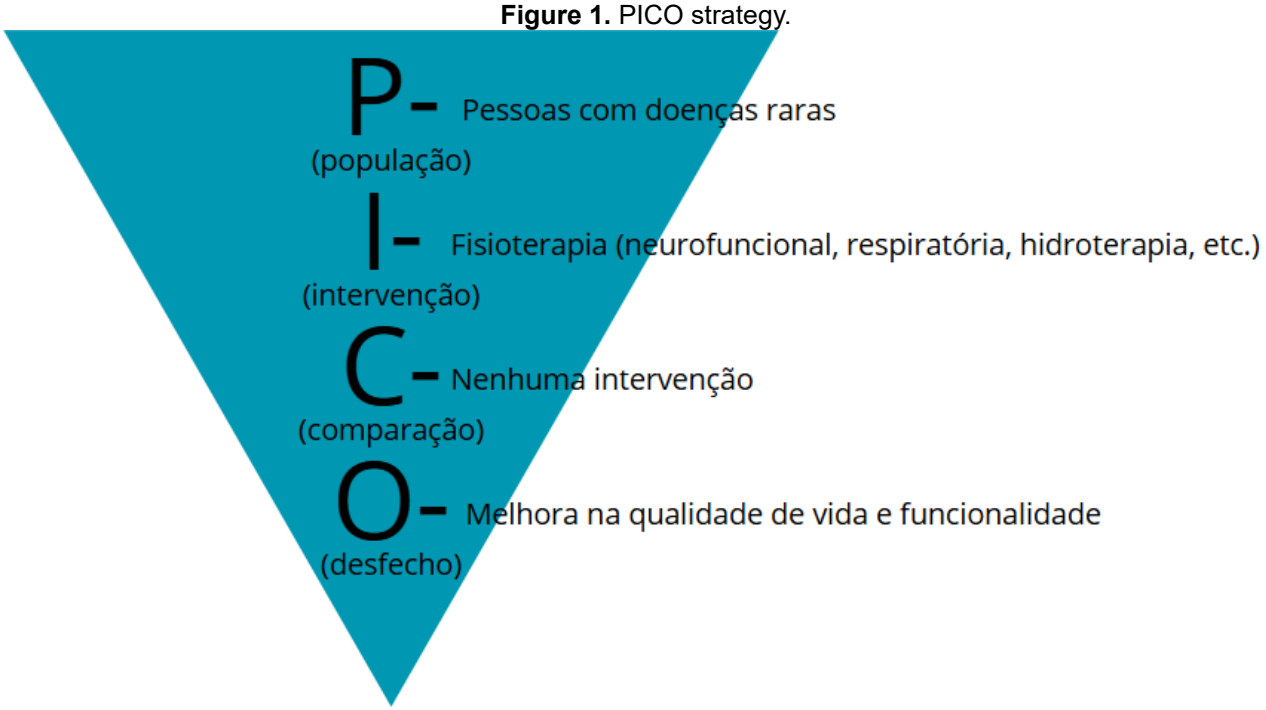
physiotherapy in any of its modalities (neurofunctional, respiratory, hydrotherapy, etc.); the comparative (C) being no intervention; and the outcome/outcome (O) being the improvement in the quality of life and functionality of the patients, as shown in Figure 1.

Figure 1. PICO strategy.



Source: The authors.

ELIGIBILITY CRITERIA

Inclusion Criteria

Studies from the last ten years, that is, between 2014 and 2024, were considered eligible. The following were selected: (1) studies involving evaluation and physical therapy intervention, in any of its modalities, in patients with rare diseases; (2) study with outcome being the evaluation of the impact of physical therapy intervention on these diseases; (3) studies that, for comparative purposes, considered other types of exercise and without a control group. There was no language restriction.

Exclusion Criteria

The following were excluded: (1) studies that investigated only other approaches to the treatment of rare diseases (medications, speech therapy, etc.); (2) studies with methodological design of narrative reviews, cohort studies, case-control, case studies and abstracts; (3) studies in which the full text was not freely available.

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SEARCH STRATEGY

A systematic search was carried out in electronic journal indexing databases: PEDro, Virtual Health Library, Cochrane and PubMed. The following descriptors related to the topic of interest were used to search: "Rare diseases"; "Physiotherapy" and its variations, using the Boolean operators "OR" and "AND" to combine the search terms (Chart 1).

Table 1. Combination of terms used for the search strategy

Study Design	Descriptors	Free Terms
Population	"Rare disease" "Rare diseases"	Not Applicable
Intervention	"Physiotherapy" "Physical Therapy"	Not Applicable

Source: Author data, 2024.

SELECTION OF STUDIES

The selection of studies was made through the Rayyan® platform. The articles obtained by the platform's search were inserted into the website, where duplicate texts were excluded. An initial evaluation and selection was carried out based on the titles and abstracts of the studies. When the initial information was not sufficient for exclusion, the full text was revised. Then, the complete studies were evaluated according to pre-defined criteria.

DATA EXTRACTION

The following data were extracted from the selected studies: 1) Author, year of publication; 2) Rare disease investigated; 3) Sample (sample size, age and gender); 4) Duration and frequency of the intervention; 5) Intervention; 6) Evaluation Instruments; 7) Outcomes.

RESULT

SELECTION OF INCLUDED STUDIES

Based on the established selection criteria, the initial search identified 20,909 articles, of which 12,962 were duplicates. After reviewing the titles and abstracts of 7,076 articles, 49 publications remained. Of these, 15 were excluded because they did not have

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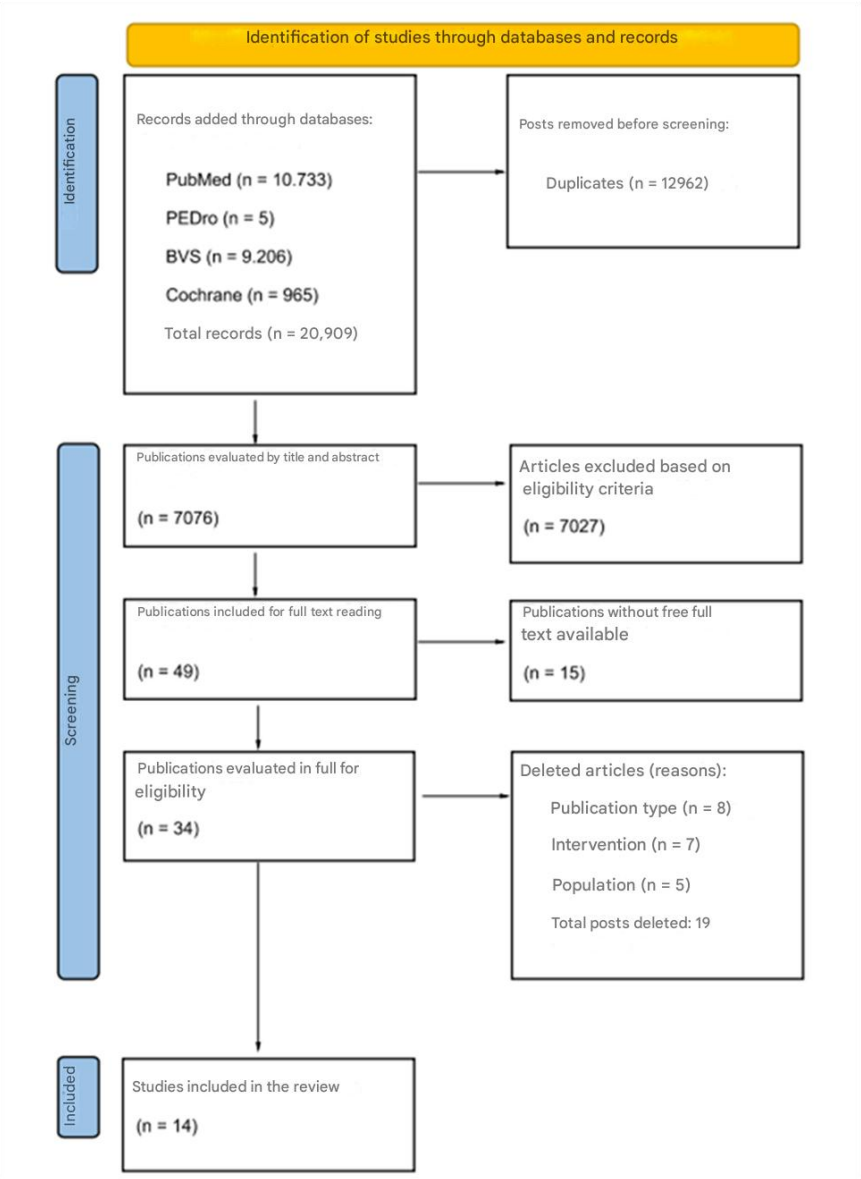
the free full text and 20 because they did not meet the eligibility criteria after reading the full text. In the end, 14 articles were considered eligible and included in the Integrative Review (Figure 2).

Figure 2. PRISMA Literature search flowchart of the included studies



Source: The authors, 2024.

DESIGNS AND SAMPLE

Of the 14 studies included in the present review, 4 are randomized controlled trials (Nakajima et al., 2021; Rannou et al. 2017; Cuesta-Barriuso, 2018; Astley, 2021), 2 are feasibility trials (Van den Berg, et al. 2015; Wittwer, Winbolt, Morris, 2019), 3 are prospective study (Su et al., 2024; El Habnouni et al., 2020; Child et al., 2023), 3 are pilot

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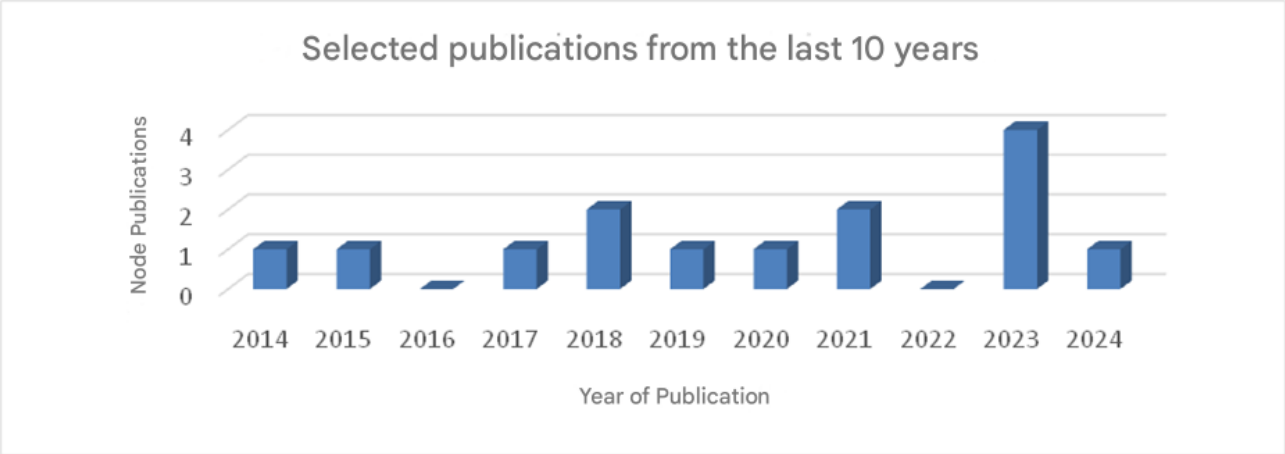
studies (Yu et al., 2023; Wallin et al., 2023; Heřmánková et al., 2023); an experimental study (Fiuza-Luces et al., 2018) and a retrospective analysis (Hoyer-Kuhn et al., 2014). Regarding the year of publication, most of the returned articles were published in 2023, as shown in Graph 1.

The rare diseases addressed in the studies were: Spinal and Bulbar Muscular Atrophy (SMA and EBMA), Muscular Dystrophy, Charcot-Marie Tooth Disease, Distal Myopathy, Sporadic Inclusion Myositis, Systemic Sclerosis/Scleroderma, Hemophilia, Mitochondrial Disease, Pompe Disease, Rett Syndrome, Takayasu's Arteritis, Congenital Myasthenic Syndrome, Duchenne Muscular Dystrophy, Multiple Sclerosis, Osteogenesis Imperfecta, Idiopathic Inflammatory Myopathy, Progressive Supranuclear Palsy, Pediatric Lymphedema, and Interstitial Lung Disease.

The total sample comprised 452 individuals, of which 130 were included in a control or conventional treatment group (Rannou et al., 2017; Cuesta-Barriuso, Gómez-Conesa, López-Pina, 2018; Astley et al., 2021; Heřmánkováet al., 2023). The other 322 participants were divided heterogeneously into the previously mentioned pathologies. In the study by Su et al. (2024), for this review, only the results of individuals with Rett Syndrome were considered.

Graph 1. Selected publications in the last 10 years.



Source: Author data, 2024.

ASSESSMENT INSTRUMENTS AND OUTCOMES

Of the 14 articles evaluated, 5 had the distance walked as an outcome, which was evaluated using the 6-Minute Walk Test (6MWT) in 2 of these studies, with the 2-Minute Walk Test (2MWT) in two others, and in only one evaluated using one-minute walking. Five studies addressed muscle strength as the outcome (primary or secondary), which was

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assessed heterogeneously between studies, and was predominantly assessed using the Manual Muscle Strength Test (3 studies). To measure Aerobic Capacity, 2 of 3 studies that addressed this outcome used a cycle ergometer with an incremental protocol, also observing respiratory parameters and heart rate (HR). In total, 5 articles brought Body Composition as the outcome, which was evaluated in 4 of them by means of Dual Energy X-ray Absorptiometry (DXA), and one through a densitometer.

Regarding Quality of Life, this was addressed in 6 of the 14 studies, using mainly the Short Form Health Survey – SF-36 scale (4 articles). Depression was the outcome in 3 studies, and each one was evaluated with a different instrument, namely: Hospital Anxiety and Depression Scale (HADS), Beck Depression Inventory-II (BDI-II) and Geriatric Depression Scale.

Because it is a heterogeneous group of diseases, and because of its rarity, many of the outcomes were specific to each pathology evaluated, using specific and validated scales and evaluation methods for the disease addressed. The evaluation instruments, as well as the key characteristics of the protocols carried out, are summarized in Chart 2.

Table 2. Publications, protocols and interventions in the RDs.

Author and Year of Publication	Rare Disease	Sample	Duration and frequency of the intervention	Intervention	Assessment Instruments	Outcomes
ASTLEY, Camila et al. (2021)	Takayasu's arteritis	N = 14 GI: 9 CG: 5 Gender: 10 female (71.4%) / 4 male (28.6%) Age: GI - 17.1 (3.72); GA - 20.4 (3.21)	3 days/week, for 12 weeks.	GI: Home exercise program with aerobic training and bodyweight strength training. Training progression every 4 weeks. GC: Conventional treatment.	Mri; Blood tests; Accelerometer; Sit and stand test; TUG; ITAS 2010; PVAS; Vasculitis Damage Index; SF-36	IL1-beta (pg/ml): CG - 2.70 (2.43); GI - 2.26 (1.27); : Δ: -1.76; p = 0.053 Passos (count/day): GC - 6,348 (3,097); GI - 12,890 (6,142); Δ: 5.463; p = 0.011; TUG (s): GC - 5.52 (0.24); GI - 5.20 (0.25); Δ: -0.50; p = 0.005; Sit and Stand (reps): CG - 16.0 (1.50); GI - 19.39 (1.36); Δ: 2.75; p = 0.012; Physical component SF-36: GC - 86.2 (61.8-94.3); GI: 85.0 (69.3-86.8); p = 0.012.

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CHILD, Claire E. et al. (2023)	Lymphoangioleiomyomatosis (LAM)	N = 15 Gender: 15 female (100%) Age: 49.0 (±7.8)	4 days/week - aerobic exercises; 3 days/week - resistance training for 12 weeks	Home exercise program with aerobic training and resistance training	TC6M; Maximal Exercise Cardiopulmonary Test; Lower limb dynamometry; Pulmonary function tests; Standardized questionnaires for fatigue and quality of life;	TC6MDistance (m): 574 ± 63; moving - 36 ± 34; p = 0.003; Predicted distance (%): 99±9; moving - 6±4; p = 0.0005; Fatigue (%): 3±6; Change: -4±5; p = 0.03.
CUESTA-BARRIUSO, Rúben; GÓMEZ-CONESA, Antonia; LÓPEZ-PINA, José-Antonio (2018)	Haemophilia	N = 27TM: 9 FE: 9 CG: 9 Age: TM: 28.0FE: 32.0GC: 37.5	TM: 2 days/week; EF: 1 time every 2 weeks for 12 weeks	TM: Manual Therapy; FE: Group therapy + home exercises	Goniometry; Perimetry; Rupture Test for patients with Hemophilia; Pain perception - VAS;	There was a change only in volunteers treated with manual therapy. Improvement in pain perception (p = 0.006), elbow flexion (p = 0.022) and arm perimeter (p = 0.050); In Av2, the improvements observed in the manual therapy group were maintained
EL HABNOUNI, Chakib et al. (2020)	Pediatric Lymphedema	N = 15 Gender: 8 male (53.3%) Age: 11.0	1 session	Manual Lymphatic Drainage	Perimetry; Calculation of the volume of members; Dermal thickness, measured by high-resolution ultrasound; Questionnaire on the usefulness and acceptability of MLD by children	Perimetry: Reduction of 4.3cm in the mean perimeter value (p = 0.024); Limb volume: Non-significant reduction of 98ml (P = 0.390); Dermal Thickness: Reduction in dermal thickness of 1.44mm (p < 0.001); Usefulness and acceptability questionnaire: Of the 12 children who responded, all reported improvement in well-being (from mild to severe) after the MLD session and found MLD useful.

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FIUZA-LUCES, Carmen et al. (2018)	Mitochondrial disease	N = 12 Gender: 8 male; 4 female Age: 46.6	3 days/week; Breathing exercises - every day in the morning and afternoon for 8 weeks	Aerobic training, strength training, and inspiratory muscle training	Cycle ergometry; Muscle strength test; Mip; TC6M; TUG; Step test; X-ray dual-energy absorptiometry (DXA); SF-36	AccelerometryThe mean levels of moderate to vigorous physical activity did not differ (P = 0.8426) between pre-training (56±20 min/week), post-training (56±21 min/week) and deconditioning (45±29 min/week). Aerobic power test: Maximum HR values (P = 0.242) and respiratory exchange ratio (P = 0.368) did not differ over time. A significant effect of time was found for all *end points* indicative of aerobic power, muscle strength, and inspiratory muscle power (all P < 0.004), except for VO2 in CPR relative to muscle mass (P = 0.006). Improvements in training were not completely lost after deconditioning (P < 0.004)ADL tests and DXA variables - virtually all patients showed the same individual response (training-induced improvement)
HEŘMÁNKOVÁ, Barbora et al. (2023)	Systemic sclerosis and idiopathic inflammatory myopathy	N = 16GI: 8 CG: 8 Gender: 16 female (100%) Age:GI: 46.5CG: 46.5	2 days/week, for 8 weeks	PFM Exercises and Physical Therapy for Musculoskeletal Problems That Limit Sexual Function	Female Sexual Function Index (FSFI), Brief Sexual Functioning Index for Women (BISF-W); Sexual Quality of Life Questionnaire - Female (SQoL-F); Health Assessment Questionnaire (HAQ); SF-36; Beck Depression Inventory-II (BDI-II).	Improvement in FSFI score by 25% in IG vs 12.5% in CG (p = 0.043); Improvement in BISF-W score by 62.5% in IG vs 12.5% in CG (p = 0.040); Improvement in HAQ of 50% in the GA vs 37.5% in the CG; Deterioration of 0% in IG vs 25% in CG (p = 0.018); Improvement in the physical component of the SF-36 of 50% in the IG vs. 37.5% in the CG (p = 0.049)

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HOYER-KUHN, Heike et al. (2014)	Osteogenesis imperfecta	N = 53 Gender: 32 male (60.4%) Age: 9.07	1st hospitalization period: 13 consecutive days of training with a one-day break; 2nd hospitalization period: 6 days; Home-based training - 6 months, 2x/day, for 6 months	Resistance training, treadmill training with body weight support, neurodevelopmental treatment, and laterally alternating body vibration (WBV).	GMFM-66 (after 12 months), Brief Motor Function Assessment (BAMF); One-minute walk; X-ray dual-energy absorptiometry (DXA);	Significant increase in motor function from Av0 to Av1 from 55.47±2.45 to 59.09±2.82 points (n=30; p<0.0001); Walking distance of 1 minute increased from Av0 to Av1 from 47.04±6.52 to 67.29±7.20 meters (n=14; p<0.0017); Significant increase in GMFM-66 score from Av0 to Av2 from 55.47±2.45 to 58.67±2.83; p=0.001; BAMF levels increased significantly from 6.84 ± 0.47 to 7.52 ± 0.41 (n = 25; p = 0.001) between Av0 and Av1;
NAKAJIMA, Takashi et al. (2021)	Rare neuromuscular disease (SMA, Spinal and bulbar muscular atrophy (SBMA), muscular dystrophy, Charcot-Marie Tooth disease, distal myopathy, sporadic inclusion myositis)	N = 24GA = 13 GB = 11 Gender: GA - 6 female (46.2%); GB - 6 female (54.5%) Age:GA - 56.6 (13.2); GB - 55.5 (7.8)	9 visits, with up to 4 visits per week, over the course of the 13 weeks. 2 blocks of 13 weeks each	Training on a treadmill with body weight support, with and without the HAL device.	TC2M; Barthel index; Manual muscle strength test; TC10M	2MWT: Compared with weight-bearing training alone, HAL training promoted a significant improvement of 10.066% in the distance covered (P = 0.037); Strength Test and 10MWT: Percentage changes in cadence in the 10MWT and in the total scores of the manual muscle strength test indicated significant improvement (P = 0.003 and P = 0.039, respectively); Barthel Index: The score showed no significant improvement in treatment 2 (with HAL) compared to treatment 1 (weight-bearing only).

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RANNOU, François et al. (2017)	Systemic sclerosis	N = 218GI = 110 CG = 108 Gender: GI - 95 female (86.4%); CG - 86 female (79.6%) Age:GI - 52.7 ± 14.8; GC - 53.1 ± 14.4.	Supervised exercise: 3x/week, 3h session, for 4 weeks 1x per day in the home exercise phase for 11 months	Hand and joint ROM exercises, aerobic training, muscle strengthening, breathing exercises, use of orthotics	Health Assessment Questionnaire Disability Index; SSc HAQ; KILL; Kapandji Index; Cochin Hand Function Scale (CHFS); Interincisal distance; EVA; SF-36; Modified Rodnan skin thickness score; Forced expiratory volume in 1s.	There was no significant difference between the groups at 12 months (p = 0.86). 1 month: HAQ ID, SSQ c HAQ and MACTAR - reduction in major disability for GI (1.13±0.61; 0.98±0.51; 13.16 7.18); Difference: -0.14; -0.14; -3.26; P = 0.0096; 0.0051; 0.0003Reduction of microstomia (p = 0.0021), improvement of global hand monility and reduction of disability and pain in the hands for the IG (Kapandji Index - p < 0.0001; CHFS - p = 0.0039). 6 months: HAQ DI - difference between groups was -0.12; p = 0.054.Microstomia - greater reduction in GI (p = 0.044); Kapandji index - improvement in hand mobility greater in GI (p = 0.048)
SU, Ting-Yu et al. (2024)	Rett Syndrome and Cerebral Palsy*	N = 6* Gender: 6 female (100%) Age: 14.5±2.9	1 day a week, for 12 weeks	Low-Intensity Extracorporeal Shock Wave Therapy in the Spastic Legs	Modified Ashworth Scale; passive ROM; Gross Motor Function Measure (GMFM-88); Muscle thickness, Acoustic Radiation Force Impulse (ARFI) and strain elastography through ultrasonography.	MAE: no statistically significant improvement in spasticity was observed (P = 0.061); Passive ROM: no statistically significant improvement was observed (P = 0.135); GMFM-88: there was an improvement in the total score (P = 0.030), but there was no statistically significant change in the variables alone (walking/running/jumping function; P = 0.156)

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VAN DEN BERG, Linda et al. (2015)	Pompe Disease	N = 23G1 = 12G2 = 11 Gender:G 1 - 7 male (58%)G2 - 5 male (45%) Age:G1 - 45.4G2 - 46.6	3 days/week 12 weeks	Eaaerobic training and Strengthening of upper limbs, lower limbs and core G1: intervention occurred at weeks 1 - 12G2: intervention occurred at weeks 13 - 24	plasma CK; Cycle ergometry with incremental protocol; HR and ventilatory parameters; Borg scale; TC6M; Core Stability; Manual dynamometry; Functional Activity (timed - 10m run; 4-step climb; rising from supine to standing); Rapid motor function test (QMFT); Body composition: mineral mass, lean mass, and body fat measured using a densitometer.	Ventilatory parameters: Wmax, VO2peak and VT improved significantly after training (P < 0.01)HR: there were no significant differences between patients' maximum HR before and after 12 weeks of training (P = 0.16); 6MWT: The mean distance walked increased by 16 m (pre: 492 ± 89; post: 508 ± 97; P = 0.01); Core StabilityThe average time they were able to remain in balance improved for all four positions (P < 0.05); Muscle Strength: Of the 9 groups evaluated, there was an increase in strength in hip flexors and shoulder abductor; Muscle Function: Reduction in time to climb 4 steps ([−0.54 to −0.04], P = 0.02) and from standing from supine to standing ([−2.0 to 0.01], p = 0.05). The QMFT score and the time to run 10m did not change.

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WALLIN, Andreas et al. (2023)	Multiple sclerosis	N = 12 Gender: 9 female (75%); 3 male (25%) Age: 54	2 to 3 days a week for 10 weeks	Group and individual balance training	Mini-BESTest; TC2M; 6 Spot Step Test (SSST); Trail Testing part B; Ray's auditory verbal learning test; Testing of modal digital symbols; Multiple Sclerosis Walking Scale - 12 (MSWS-12); Falls Efficacy Scale (FES-I); Frenchay Activity Indices; Multiple Sclerosis Impact Scale-29; Modified Fatigue Impact Scale; Hospital Anxiety and Depression Scale; EuroQol-5D; Life satisfaction list; Chronic Health Conditions Acceptance Scale.	Mini-BESTest: Positive effect trend evaluated, with a median difference between baseline and follow-up assessments of 3.5 points (3.5; − 1 to 7); TC10m: Positive trend, increase in maximum gait speed, indicating improvement in walking (0.04; − 0.04 to 0.25); slightly negative trend for the same test with the self-selected gait speed (− 0.04; − 0.17 to 0.23); 2MWT: Negative trend, shorter distance covered in the follow-up assessment (− 10.2; − 14.4 to 8.4); SSST and MSWS-12: Positive trend in both, indicating improvement in complex walking (− 1.7; − 3.5 to 3.5) and in the way participants perceived the impact of MS on walking ability (− 5.5; − 16 to 1); FES-I: Negative trend, indicating greater concern about falls (1.5; − 8 to 7).
WITTWER, Joanne E.; WINBOLT, Margaret; MORRIS, Meg E. (2019)	Progressive Supranuclear Palsy	N = 5 Gender: 3 female (60%); 2 male (40%) Age: 54-74 years	2 days/week8 sessions for 4 weeks	Music-guided gait training	Addenbrooke's Cognitive Examination-III (ACE-III); Geriatric depression scale; Personal musical preference scale; Evaluation of the physiological profile. PSPRS and UPDRS; Spatiotemporal gait measurements (GAITRite® mat).	Geriatric Depression Scale: Four of the five participants had scores suggestive of depression. On Av1 the scores remained largely unchanged; No participant showed improvement in all post-intervention measures; 60% increased walking speed at a "comfortable pace" after the intervention; 80% of participants improved knee extensor strength, although only one participant achieved normal values.

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YU, Michael Kwan Leung et al. (2023)	Hereditary neuromuscular disorder (Spinal Muscular Atrophy (SMA), Congenital Myasthenic Syndrome (CMS), and Duchenne Muscular Dystrophy (DMD)).	N = 8	3 days/week for 16 weeks	Telerehabilitation - breathing exercises and Strengthening of upper limbs and lower limbs	Spirometry (FVC, PiMax, PeMax, maximal cough flow); Manual muscle strength test; Hydraulic manual dynamometer; Jamar clamp meter; TC6M; Pediatric Quality of Life Inventory 3.0 Neuromuscular Module survey; User satisfaction survey; Borg scale.	Borg scale: All volunteers rated the Borg scale as ≤13; Spirometry: MIP improved after the study period (pre: 35.0; post: 47.5; P = 0.028); Other scales, including MEP, PCF, hand and pinch strength, and walking distance, remained stable after the study period, but not statistically significant results. Pediatric Quality of Life Inventory 3.0 Neuromuscular Module survey: volunteers reported improvements in health-related quality of life after the study period (total score - pre: 74.5; post: 87.0; P = 0.036)

PHYSIOTHERAPEUTIC INTERVENTIONS AND NUMBER OF SESSIONS

The duration of the reviewed studies ranged from 8 to 13 weeks, with variance from 1 session every 15 days to 4 sessions per week, in addition to daily exercises to be done at home. The sessions varied in time between 40 minutes and 90 minutes and the home exercises varied between 20 and 30 minutes; Only 6 studies performed follow-up after completion of the protocol (follow-up).

Regarding the type of approach used, 10 studies performed Motor Physical Therapy, 3 addressed Respiratory Physical Therapy, always associated with another modality (Motor or Cardiovascular). Manual Therapy (Manual Lymphatic Drainage) was used in one of the studies, and Neurofunctional Physical Therapy was applied in 2 of the 14 articles evaluated. One of the protocols involved Physical Therapy in Women's Health.

MAIN RESULTS

Quality of Life

Physical therapy promoted improvement in the Physical Component of the SF-36 in two studies (Astley et al., 2021; Heřmánková et al., 2023). The study by Fiuza-Luces (2018), which also used this questionnaire as a way to assess quality of life, promoted a statistically significant improvement in the "General Health Status" component of the

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questionnaire (P = 0.001). Another article that also used the SF-36 as an evaluative method was that of Rannou et al. (2017), and there was no statistically significant difference in any of the components of the questionnaire for either of the two groups (IG and CG). Scores varied little between evaluations.

Child et al. (2023) used the Visual Analogue Scale (VAS) and the ATAQ-LAM (A Tool to Assess Quality of Life in LAM) as indicators of quality of life, an aggregation of subscales specific to physical health. Both scales showed improvement with treatment (P = 0.009 and P = 0.03, respectively).

In the study by Yu and colleagues (2023), quality of life was assessed by the Pediatric Quality of Life Inventory 3.0 Neuromuscular Module survey, and volunteers reported improvements in health-related quality of life after the study period (pre-intervention total score: 74.5; post: 87.0; P = 0.036).

DISCUSSION

This study sought to critically analyze the available scientific evidence on the physiotherapeutic approach in the evaluation and treatment of rare diseases. The search was carried out to include a wide variety of conditions, with multiple system involvements.

Initially, the research aimed to find only Clinical Trials and Experimental Studies, following the PRISMA guidelines. However, due to the low number of evidences, it was decided to expand the search to other types of publications. Although the Ministry of Health recommends that these diseases usually do not have a treatment that leads to a cure, and that rehabilitation services are alternatives to mitigate symptoms, there is still a lack of research and investments for the expansion of this area, but it can be observed that the number of publications on the subject has increased when compared to previous years (BRASIL, 2022).

The diseases addressed were of different etiologies, most of which were of a neurological nature. In addition, three articles address autoimmune diseases. Both in the forms of evaluation and intervention, the difficulty and non-standardization of effective conducts for this public is verified. The analyzed publications presented many divergent conducts, in addition to several specific evaluation questionnaires, making it difficult to compare results between them.

One of the outcomes expected in this review was the assessment of quality of life. Although directed towards this, many studies have not evaluated it through validated

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questionnaires. In articles that used validated assessment instruments, the intervention groups showed regular and/or good quality of life. However, quality of life is a multifactorial and subjective perception, which depends on the context in which the individual is inserted, his goals and expectations. In all the articles analyzed in this review, there was an improvement in some characteristic due to physical therapy interventions, such as strength gain, improvement in gait speed and cadence, and reduction of disabilities, which can be considered as an improvement in quality of life.

In the study by Haraldstad and colleagues (2019), it is described that medical advances have reflected in the increase in the number of people living with chronic diseases. The change in survival of this population has generated greater interest in research on quality of life, which has been frequently evaluated in studies, highlighting the importance of considering not only medical outcomes, but also the impact of treatment on patients' lives when evaluating the advantages of different treatment options.

As adverse effects, a part of the volunteers reported muscle pain, especially in studies in which the protocol involves strength training (Van Den Berg et al., 2015; Nakajima et al., 2021). In addition, there were no other complications, and we can conclude that the initial evidence suggests that physical therapy intervention is a safe treatment for this population.

In longer studies, with evaluations far from the initial one, there was a sample loss, with only part of the volunteers returning for follow-up (Hoyer-Kuhn et al., 2014; Rannou et al., 2017). In addition, in the study by Rannou et al. (2017), it was observed that the beneficial effects of the therapies were not maintained in the long term. This may occur due to the progressive and degenerative nature of most rare diseases, in which many have no cure, so physiotherapy alone does not have the ability to prevent the natural progression of the pathology, only to slow down the speed of its advancement and attenuate the symptoms.

In the study by Su et al. (2024), shockwave therapy was used to reduce spasticity in children with Rett Syndrome and Cerebral Palsy. For most of the parameters evaluated, a statistically significant improvement was observed only for children with CP, and one of the hypotheses raised for this phenomenon is the progressive aspect of Rett's disease, when compared to CP. Being a neurodevelopmental disorder, patients with Rett syndrome have a regressive course in motor functions, spasticity, and muscle stiffness over time.

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The study by Rannou and colleagues (2017) showed low adherence to home exercises among volunteers, unlike the study by Yu et al. (2023) and Child et al. (2023), in which there was good adherence to the protocol. Individuals were more willing to adhere to telerehabilitation compared to in-person rehabilitation, and recommended remote practice as a standard of care for neuromuscular and other diseases (Yu et al., 2023). One hypothesis for this discrepancy is the Covid-19 pandemic, which boosted teleconsultations.

In a study on the use of telemedicine during the pandemic, the authors reported that frequent use of telemedicine was identified as promising. The COVID-19 pandemic has accelerated the regulation of this practice in several countries, as it is low cost and well accepted, and has demonstrated that telemedicine can be an additional and useful tool in services that would be absent if this technology did not exist, offering new opportunities to support health systems and human capacities (Scheffer et al., 2022).

Although only a small portion of the studies contained in this review are Clinical Trials (Astley et al., 2021; Cuesta-Barriuso, Gómez-Conesa, López-Pina, 2018; Nakajima et al., 2021; Rannou et al., 2017), all the articles analyzed brought the structure of the protocol carried out.

The transparency of the protocols has several advantages, such as contributing to advances in clinical practice and avoiding waste of resources. The prior disclosure of these protocols allows the scientific community, health professionals, and patients to follow the progress of the research, and facilitates the replication of the program in other studies or by professionals in the field. In addition, authors benefit by promoting their research groups, establishing new collaborations, contributing intellectually to the field, and increasing their academic output (Hazime and Cardoso, 2021).

Regarding functionality, no study directly addressed it as an outcome. However, it is known that this aspect takes into account bodily functions, activity and participation, and environmental factors. The study by Wittwer, Winbolt, and Morris (2019) exemplifies this relationship well, as the participant with the lowest score on the Geriatric Depression Scale (higher scores suggest the presence of depressive symptoms) was the most independent participant in terms of community and social participation. It was possible to observe the improvement of aspects related to functionality in several articles analyzed, such as increase in distance traveled, improvement in pain perception, improvement induced by ADL training, evolution in sexual function and motor function, among other outcomes (Table 2) that may provide greater interaction and participation of these individuals in society.

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According to Di Nubila (2010), the simple "diagnosis" alone does not anticipate the demand for services, hospitalizations, the level of care or functional results. In addition, the presence of a disease or disorder is not a reliable indicator of social integration. This implies that if we rely solely on the medical classification of diagnoses, we will not have enough information for health management and planning purposes.

Souza, Silva and Carvalho (2010) state that the diversity of sources in reviews, together with the multiple objectives, aims to provide a consistent and understandable view of complex concepts, theories or relevant health problems. Thus, the results of this review are relevant, since, to the author's knowledge, no other studies were found that addressed the role of physical therapy in rare diseases in a global way, considering all its modalities in pathologies of the most diverse etiologies.

CONCLUSION

Physiotherapy, regardless of the modality practiced, promotes positive effects on the quality of life and functionality of people with rare diseases. However, these impacts do not last for long periods due to their degenerative and progressive character. However, knowing that functionality involves activity and participation, and that this directly impacts the individual's quality of life, physiotherapy is a good treatment option to attenuate and delay the progression of rare disease symptoms, especially in the motor sphere.

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