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A systematic review of telemedicine for neuromuscular diseases: components and determinants of practice

Deniz Senyel[1,2*], Katja Senn[1], James Boyd[2] and Klaus Nagels[1]

Abstract

Introduction Neuromuscular diseases (NMDs) entail a group of mostly inherited genetic disorders with heterogeneous phenotypes impacting muscles, the central or peripheral nervous system. They can lead to severe disabilities and shortened lifespans. Despite their severity, NMDs often lack in public awareness and appropriate medical and social support. Telemedicine can improve patients' and caregivers' lives by enhancing continuity of and access to care. The first aim of this systematic review was to summarise the status quo of telemedicine services for patients with NMDs. Secondly, barriers and facilitators of the respective implementation processes should be analysed.

Methods The databases PubMed, Web of Science and CENTRAL by Cochrane were searched in May 2022. To be truly explorative, any original evidence from any setting was included. Two independent researchers completed the screening process. Data was extracted and analysed using the taxonomy of Bashshur et al. (2011) and the Consolidated Framework for Implementation Research (CFIR).

Results Fifty-seven original papers were included in the systematic review. The results showed a high representation of teleconsultations and remote monitoring studies. Teleconsultations replaced in person appointments and telemonitoring mostly focused on ventilation. Physical therapy, pulmonology, neurology, and psychology were the most represented medical specialties. We found barriers and facilitators relating to implementation mainly referred to the intervention and the individuals involved. Technical errors and inaccessibility due to a lack of technical devices or the patient's disability were stated as hindrances. A positive mindset of users as well as patient empowerment were necessary for the adoption of new technology. Technophobia or uncertainty around technology negatively impacted the implementation process.

Discussion This systematic review provides an overview of the current use of telemedicine in patients with NMDs. The distribution of telemedicine interventions between the defined domains was very heterogenous. Previous research has neglected to fully describe the implementation process of telemedicine for NMDs.

Conclusion The evidence shows that telemedicine can benefit patients with NMDs in a multitude of ways. Therefore, health policies should endorse and incentivise the uptake of telemedicine by institutions and health care workers. Further research needs to be conducted to confirm the current evidence and close existing research gaps.

*Correspondence:
Deniz Senyel
d.senyel@latrobe.edu.au
Full list of author information is available at the end of the article

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Introduction

Neuromuscular diseases (NMDs) are a heterogeneous group of disorders, that affect the nerves controlling muscles, leading to muscle weakness, wasting, and other related symptoms [1]. NMDs are often hereditary and have been linked to 500 different affected genes [2, 3]. Most NMDs are classified as rare diseases. The prevalence of NMDs can vary widely and, even for common diagnostic groups, the prevalence ranges between 0.1 to 60 per 100,000 [4]. The onset, cause, and course of the disease vary widely between disorders [5]. While each individual's experience is unique, there are common disability-related challenges faced by patients with NMDs. Acknowledging these commonalities and addressing the unique needs of each person are essential for providing comprehensive care and support to individuals and their families living with NMDs. NMDs are highly complex diseases defined by a degenerative course and progressive muscle weakness as the main symptom. Their impact extends beyond the musculoskeletal system, affecting various organs and systems throughout the body, such as eyes, lungs or the brain [1, 2]. As a result, patients suffer from a reduced quality of life and a significant disease burden [2, 6]. Multidisciplinary care is often considered the optimal approach for providing holistic treatment and symptomatic management for individuals with NMDs [7–11]. The needs of patients during disease progression are ever changing based on disease stage, symptom burden, and personal priorities. General practitioners, specialists, and allied health professionals each bring unique expertise to the care team, allowing for comprehensive, patient-centred care that adapts to changing needs and priorities throughout the course of the disease and ensures continuity and quality of care [1, 12, 13]. Recognising and supporting caregivers is crucial in the care of NMD patients. Most NMD patients receive informal care, often provided by their partner or family members. The caregiver burden increases with the progression of the patient's disease. In severe cases, it can lead to psychological distress and burnout, a state of physical and emotional exhaustion [14–17].

Mobile health apps, teleconsultation and telemonitoring have been proven to be useful tools in the management and treatment of chronic diseases such as diabetes, heart failure, asthma, chronic obstructive pulmonary disease, and cancer. They have the potential to increase treatment adherence, support self-management, and promote continuity of care [18–20]. They have the potential to reduce hospital admissions, decrease mortality rates, and lessen health services usage [21–24]. The research focus in telemedicine for NMDs varies between disorders. A recent systematic review by Helleman et al. showed telemedicine for ALS patients to be a useful option for remote monitoring, consultations, and follow-ups [25]. From a patient's perspective it can be time- and cost-saving while reducing stress and fatigue. While telemedicine has demonstrated its value in certain NMDs like ALS, its usage in the care of other NMDs have not been as extensively studied or described.

This systematic review aims to identify telemedicine interventions for patients with NMDs and analyse the barriers and facilitators of the implementation process associated with telemedicine for NMD patients. The taxonomy by Bashshur et al. will be used to standardise terminology and make it easier to categorise and study the various telehealth interventions and services [26]. The term "Telemedicine" will be used as an umbrella term to encompass a broad range of remote healthcare services and technologies. This is done to avoid the potential ambiguities and unclarities that can arise from newer terms like "e-health" or "telehealth". This review will provide an overview of the status quo and will offer recommendations for future innovations.

Methods

This systematic review followed the PRISMA [27] checklist. The study protocol was registered on PROSPERO (ID: CRD42022325481).

Databases and search strategy

For the literature search PubMed, Web of Science, and the Cochrane database CENTRAL were used as sources. If full text could not be found, the authors were contacted. The final search was conducted in May of 2022.

The search strategy consisted of two major themes: Firstly, synonyms for NMDs and secondly, synonyms and subcategories for telemedicine. The full search strings can be found in the supplementary file 1.

Study selection

The study selection was conducted by two reviewers KS and DS. The following inclusion criteria were applied: Studies from any country with any healthcare and insurance system were eligible to maximise the diversity and inclusivity of the evidence base. No restrictions regarding cultural or socio-economic context were made to be truly explorative. Articles were eligible for inclusion if their study population consisted of patients with one or

more types of NMDs. Since a single comprehensive list of all NMDs could not be found, the list of NMDs by the Muscular Dystrophy Association (MDA) was used as a reference [28]. If a disease could not be found under the listed disorders, the International Classification of Diseases (ICD) was consulted [29]. No limitations regarding sex, age, race, or nationality were made. All types of telemedicine were eligible for inclusion. The taxonomy by Bashshur et al. was used as a guiding definition [26]. Bashshur uses telemedicine in his paper as the original term for ICT in healthcare. The domains include the following components:

- *Telehealth*: Health behavior & education; Health & disease epidemiology; Environmental/Industrial health; Health management & policy.
- *E-health*: Electronic health record; Health information; Clinical decision support system; Physician order entry.
- *M-health*: Clinical support; Health worker support; Remote data collection; Helplines.

Interventions could be implemented on a national, communal, or institutional level. The users could include patients, caregivers, and healthcare workers. Only primary research was included. Due to the explorative nature of the systematic review, no major restrictions regarding study types were made. Only articles written in English or German were included. Due to the rapid pace of technological progress, only studies from the last ten years were considered. This ensured that the telemedicine interventions were not out-of-date or obsolete.

Studies were excluded if no specific diagnostic group was mentioned. Further reviews, study protocols and commentaries were excluded.

Data extraction and analysis

The data extraction and analysis were done by DS. From the included studies the following data points were extracted: authors, year of publication, country, included NMDs, intervention type and analysed outcomes. Additionally, barriers and facilitators of the implementation process were collected. The Consolidated Framework for Implementation Research (CFIR) was used to guide the extraction process [30]. The CFIR is an established framework for the analysis of implementation processes. Based on this structure, a detailed coding manual with operationalised definitions for each construct was created. This manual served as a reference guide to ensure that the extraction and coding process was systematic and reproducible.

The data synthesis was done narratively. Since no effect measures were used, a quantitative analysis was not applicable. Firstly, the types of telemedicine interventions were clustered according to the domains described by Bashshur et al., to gain a comprehensive understanding of the current landscape of telemedicine applications [26]. Secondly, the CFIR was used to label quotes on implementation barriers and facilitators [30].

No meta-analysis was conducted as there are no quantitative outcomes to analyse. Further, the heterogeneity of the studies was not assessed. Due to the broad inclusion criteria, a high heterogeneity could be expected. Since the focus of this systematic review lies on the intervention types, rather than on their effectiveness, subgroup analyses were not performed. Equally no sensitivity analyses were conducted. The focus of the systematic review was not to summarise evidence regarding a specific intervention, it was an exploration of the current telemedicine options for patients with NMDs.

Risk of bias

The study protocol stated a risk of bias assessment using the RoB 2 and ROBINS-I tools [31, 32]. This was later changed to the JBI's critical appraisal tools as they offered a wider selection of checklists [33]. No meta-bias was analyzed since the outcomes of the studies were not a point of interest.

Results

Included studies

Figure 1 depicts the study selection process for the systematic review, including a total of 57 reports. These included four report pairs with interlinked content. Ando et al. published two papers on the Intervention Careportal in 2019 and 2021 [34, 35]. Hobson et al. conducted one study with results disseminated across two publications [36, 37]. Martinet et al. conducted two studies utilising the same intervention but with distinct comparison groups and study populations [38, 39]. Lastly, Sobierajska-Rek et al. and Wasilewska et al. published two articles addressing different subsections of one main study [40, 41]. Studies excluded during the full text screening process can be found in supplementary file 2.

Study characteristics

Table 1 presents an overview of the study characteristics. A total of 25 studies were carried out using a cross-sectional design [34, 35, 40, 42–63]. Additionally, the review included two case series [64, 65] and one case–control study [66]. Among the studies, 16 adopted a cohort study design [41, 67–81], while ten employed an experimental design [36–39, 82–87]. The remaining three reports were method papers [88–90]. Geographically, the majority of the studies took place in Europe [34–43, 47–49, 53, 58–60, 62, 65, 66, 68, 69, 72–74, 76, 80–82, 84, 87–90] and

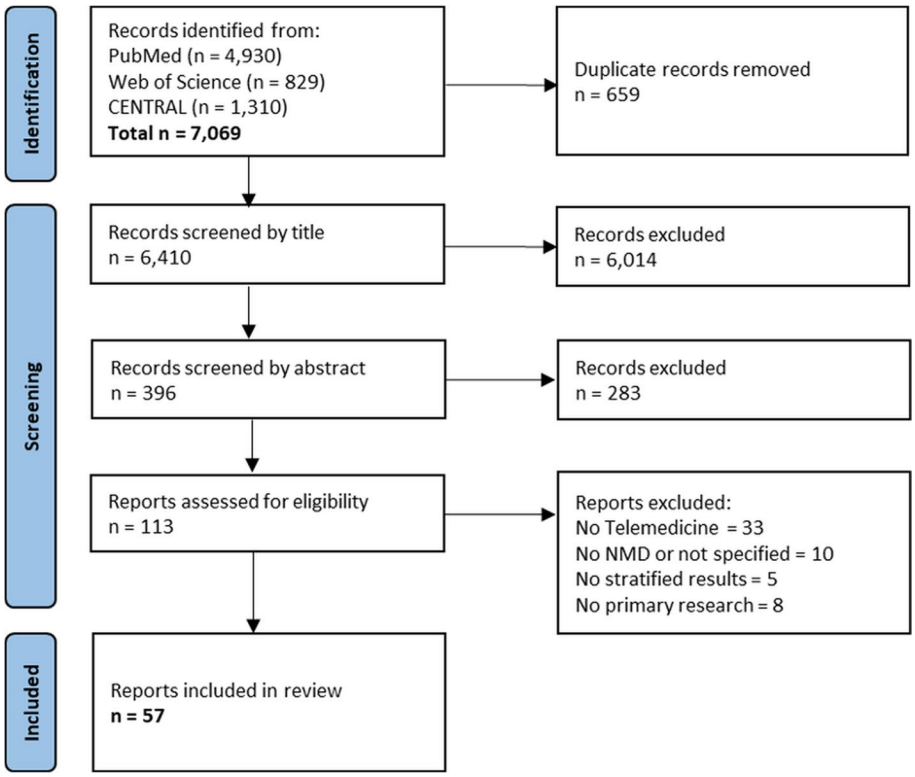

Fig. 1 Flow diagram of the identified studies (Source: own depiction)

the USA [44–46, 51, 52, 54, 56, 57, 63, 67, 71, 77–79, 85, 86]. Two studies were conducted in Canada [50, 64] and one in each of the following countries: Japan [70], India [75], Brazil [83], and Australia [55]. One study included participants from around the globe [61].

A total of twenty-nine studies exclusively focussed on ALS patients [34–37, 42, 46, 48, 51, 52, 55–57, 62, 69–73, 76–81, 85–90], while another four studies included ALS patients alongside other NMD diagnostic groups [53, 54, 58, 60]. The study outcomes assessed in these studies varied widely. Clinical outcomes, such as physical and cognitive function, as well as mental health, were often used. Further, user satisfaction and utilisation measurements were applied to evaluate interventions. For patient registry studies, epidemiological statistics, including prevalence, were commonly employed as outcome measures.

Risk of bias

While the primary focus has been on exploring the availability of telemedicine interventions for patients with NMDS, it is crucial not to overlook the evaluation of individual study quality and the potential impact of bias. In summary, most studies demonstrated a low risk of bias and employed sound methods and procedures. However, certain limitations, such as the lack of comparison groups, insufficient follow up time, and some inadequate reporting, should be noted. Visual depictions and the complete analysis can be found in supplementary material 3. Three reports were not assessed as they only presented a method paper without empirical results [88–90].

Telemedicine domains of included interventions

In the following sections the telemedicine interventions included in the analysis will be examined, guided by the taxonomy by Bashshur et al [26]. According to their definition, telemedicine comprises of three major domains: *telehealth, e-health, and m-health*. Eight studies were categorised under the telehealth domain, encompassing all traditional public health areas. *E-health,* mainly describing the online storage of information and supporting tools for physicians, was represented by ten studies. The majority of studies fell within the *m-health* domain, a rapidly growing field that leverages mobile devices like smartphones and tablets to deliver healthcare services, monitor patients remotely, and support self-management. Given that interventions could encompass elements from different domains, multiple mentions or references to different domains is possible. As stated, there were instances where multiple reports featured identical telemedicine interventions [34–39]. In order not to bias the results, identical interventions were counted as one during the analysis of the telemedicine

Table 1 Study characteristics

Author	Year	Study type	Country	NMD[a]	Intervention	Outcome
Abdulla et al. [42]	2014	Cross-sectional	Germany	ALS	Not applicable	Information needs and information seeking behaviour
Alexanderson et al. [82]	2014	RCT	Sweden	PM; DM	Home exercise with tele-support	Physical function
Ambrosini et al. [43]	2018	Cross-sectional	Italy	DMD; BMD; SMA; CMT; MGSD; SBMA; TTR-FAP	Patient registry	Epidemiology
Ando et al. [35]	2019	Delphi + cross-sectional	England	ALS	Careportal: a telehealth communication device	Utilisation Physical function
Ando et al. [34]	2021	Cross-sectional	England	ALS	Careportal: a telehealth communication device	Satisfaction
Anil et al. [44]	2020	Cross-sectional	USA	MG	Patient registry	Epidemiology
Astley et al. [83]	2021	Before-after	Brazil	JDM	Home exercise program	Mental Health QoL Physical function
Bankole et al. [84]	2016	Cross-over	France	FSHD	Home exercise program	Physical function
Benjamin et al. [45]	2019	Cross-sectional	USA	Not applicable	Blended curricula on physical examination in patients with NMDs	Knowledge Score
Berry et al. [46]	2019	Cross-sectional	USA	ALS	Remote data collection via the Beiwe smartphone app	Physical function Cognitive function
Bettio et al. [47]	2021	Cross-sectional	Italy	FSHD	Patient registry	Epidemiology
Capozzo et al. [48]	2020	Cross-sectional	Italy	ALS	Teleconsultation	Satisfaction Physical function
Cesareo et al. [49]	2020	Cross-sectional	Italy	DMD; LGMD	Wearable device for pulmonary monitoring	Physical function Utilisation
Christodoulou et al. [85]	2016	Cross-over	USA	ALS	Telephone based cognitive-behavioural assessment	Cognitive function
Climans et al. [50]	2017	Cross-sectional	Canada	MyD	Not applicable	Computer access and use
Contesse et al. [67]	2021	Cohort study	USA	DMD	Duchenne video assessment scorecards	Physical function
Garuti et al. [68]	2013	Cohort study	Italy	PPS; BMD; DMD; MD; SMA; MFM; PD	Respicard: remote pulmonary monitoring	Utilisation Hospitalisation rates
Geronimo et al. [52]	2017	Cross-sectional	USA	ALS	Teleconsultation	Satisfaction
Geronimo et al. [51]	2019	Cross-sectional	USA	ALS	Remote pulmonary function testing	Physical function Utilisation
Grigull et al. [53]	2016	Cross-sectional	Germany	MD; PD; SMA; ALS	Diagnostic support tool via questionnaire	Accuracy
Helleman et al. [69]	2020	Cohort study	Netherlands	ALS	App-based home monitoring and coaching	Satisfaction Utilisation
Hobson et al. [36]	2019a	RCT	England	ALS	App-based home monitoring and coaching	Mental Health QoL Satisfaction Adverse events
Hobson et al. [37]	2019b	RCT	England	ALS	App-based home monitoring and coaching	Utilisation
Hooshmand et al. [54]	2021	Cross-sectional	USA	MG; ALS; CMT; BMD; MyD; LEMS	Teleconsultation	Satisfaction
James et al. [55]	2019	Cross-sectional	Australia	ALS	Teleconsultation	Physical function Satisfaction

Table 1 (continued)

Author	Year	Study type	Country	NMD[a]	Intervention	Outcome
Kamei et al. [70]	2018	Cohort study	Japan	ALS	Telenursing/Telemonitoring via app	Physical function Utilisation Satisfaction
Levi et al. [71]	2017	Cohort study	USA	ALS	Making Your Wishes Known—Decision support tool for patients	Physical function Mental Health QoL Satisfaction Decisional Conflict Time and Effort
Longinetti et al. [72]	2018	Cohort study	Sweden	ALS	Patient registry	Epidemiology
Malek et al. [56]	2014	Cross-sectional	USA	ALS	Patient registry	Epidemiology
Marchi et al. [73]	2021	Cohort study	Italy	ALS	Teleconsultation	Physical function Satisfaction
Martinez et al. [39]	2014	Before-after	Spain	MG; BMD; LGMD	Online Cognitive-Behavioural Therapy	QoL
Martinez et al. [38]	2021	Before-after	Spain	MG; FSHD; BMD; LGMD; EDMD; SMA; CMT; DM; HSP	Online Cognitive-Behavioural Therapy	QoL
McErlane et al. [74]	2021	Cohort study	England	DMD	Wrist-worn wearable device (accelerometer) + App	Physical function
Menon et al. [64]	2021	Case series	Canada	MG	Teleconsultation	Virtual MG index vs. in-person equivalent
Naveen et al. [75]	2020	Cohort study	India	DM; OM; ASS; JDM; PM; NM	Teleconsultation	Adverse effects Utilisation
Newton et al. [76]	2020	Cohort study	Scotland	ALS	Teleconsultation	Physical function Satisfaction
Paganoni et al. [77]	2019	Cohort study	USA	ALS	Teleconsultation	Cost
Palumbo et al. [88]	2021	Not applicable	Italy	ALS	Telemonitoring/-consultation via app SIMPLE	Not applicable
Portaro et al. [65]	2017	Case series	Italy	FSHD	Telemonitoring via app + Teleconsultation and rehabilitation	Physical function Mental Health Satisfaction
Pulley et al. [57]	2019	Cross-sectional	USA	ALS	Asynchronous videorecorded assessment	Satisfaction
Ricci et al. [89]	2018	Not applicable	Italy	PD	AIGkit: App for patients with PD	Not applicable
Roman et al. [78]	2021	Cohort study	USA	ALS	Teleconsultation regarding communication devices	Satisfaction
Roy et al. [58]	2015	Cross-sectional	Belgium	62 NMDS	Patient registry	Epidemiology
Scalvini et al. [90]	2018	Not applicable	Italy	ALS	Telemonitoring / -consultation /-rehabilitation	Not applicable
Selkirk et al. [79]	2017	Cohort study	USA	ALS	Teleconsultation	Quality of Care Physical function
Sobierajska-Rek et al. [40]	2020	Cross-sectional	Poland	DMD	Telerehabilitation via blended home exercise program + motor assessment tool via app	Utilisation
Sobierajska-Rek et al. [59]	2021	Cross-sectional	Poland	DMD	Telerehabilitation via home respiratory exercise	Satisfaction Utilisation
Spiliopoulos et al. [60]	2022	Cross-sectional	Greece	MP; MG; ALS	Teleconsultation	Number of consultations
Tawfik et al. [61]	2021	Cross-sectional	International	Not applicable	Virtual neuromuscular ultrasound courses	Utilisation Satisfaction

Table 1 (continued)

Author	Year	Study type	Country	NMD[a]	Intervention	Outcome
Trucco et al. [66]	2019	Case–control	Italy	DMD; SMA; CM; LGMD; CMD	Telemonitoring	Hospitalisation Satisfaction Physical function Utilisation Caregiver burden
van Eijk et al. [80]	2019	Cohort study	Netherlands	ALS	Telemonitoring using Accelerometer	Physical function
Vasta et al. [62]	2021	Cross-sectional	Italy	ALS	Teleconsultation	Satisfaction
Vitacca et al. [81]	2012	Cohort study	Italy	ALS	Teleconsultation; Telemonitoring	Utilisation Cost Complexity of Management and Burden of Care
Wasilewska et al. [41]	2021	Cohort study	Poland	DMD	AioCare System: Telemonitoring of Pulmonary Function	Physical function Satisfaction
White et al. [63]	2019	Cross-sectional	USA	DMD	DMD video assessment	Utilisation Satisfaction
Wills et al. [86]	2019	RCT	USA	ALS	App for nutritional counselling	Utilisation Adverse events Physical function
Wit et al. [87]	2020	RCT	Netherlands	ALS	Blended psychological support system for caregivers	Mental health Caregiver burden QoL Self-efficacy Satisfaction

[a] *ALS* Amyotrophic Lateral Sclerosis, *ASS* Anti-Synthetase Syndrome, *BMD* Becker Muscular Dystrophy, *CM* Congenital Myopathy, *CMD* Congenital Muscular Dystrophy, *CMT* Charcot-Marie-Tooth Disease, *DM* Dermatomyositis, *DMD* Duchenne Muscular Dystrophy, *EDMD* Emery-Dreifuss Muscular Dystrophy, *FSHD* Facioscapulohumeral Muscular Dystrophy, *HSP* Hereditary Spastic Paraparesis, *JDM* Juvenile Dermatomyositis, *LEMS* Lambert-Eaton-Myasthenic-Syndrome, *LGMD* Limb-Girdle Muscular Dystrophy, *MyD* Myotonic Dystrophy, *MD* Muscular Dystrophy, *MFM* Myofibrillar Myopathies, *MG* Myasthenia Gravis, *MGSD* Muscle glycogenosis, *MP* Myopathy, *NM* Necrotizing Myositis, *OM* Overlap Myositis, *PD* Pompe Disease, *PM* Polymyositis, *PPS* Post-Polio Syndrome, *SBMA* Spinal and Bulbar Muscular Atrophy, *SMA* Spinal Muscular Atrophy, *TTR-FAP* Transthyretin Familial Amyloid Polyneuropathy

domains and components. The distribution of telemedicine domains is illustrated in Fig. 2a.

Telehealth

The studies within the *telehealth* domain were mostly epidemiological studies. Six studies described online patient registries for one or more NMDs [43, 44, 47, 56, 58, 72]. The remaining two studies were categorised under health education. One study introduced a blended curriculum focusing on physical examinations for patients with NMDs [45] while another detailed a virtual neuromuscular ultrasound course [61]. The distribution of the *telehealth* domain can be seen in Fig. 2b.

E-Health

The second smallest domain was *e-health* (Fig. 3). Within this domain, three studies incorporated electronic health records [69, 89, 90]. Health information was the subject of five studies, with two of these not providing an intervention but instead investigating patients' computer use and information seeking behaviour [42, 50]. Only two interventions described clinical

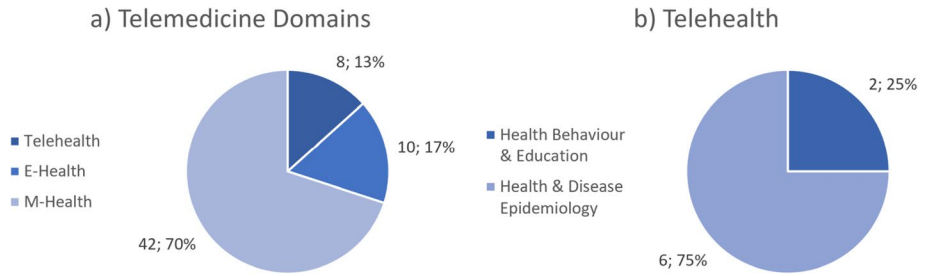

Fig. 2 Distribution of the telemedicine (**a**) and telehealth domains (**b**) (Source: own depiction)

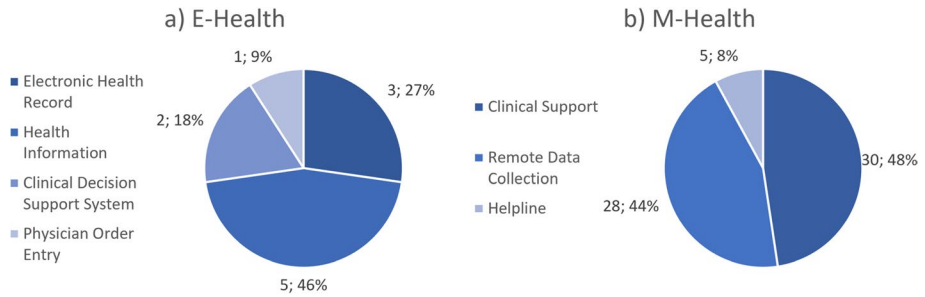

Fig. 3 Distribution of the e-Health (**a**) and m-health domains (**b**) (Source: own depiction)

decision support systems, one supporting physicians during the diagnostic phase [53] and another supporting patients with advanced care planning [71]. A singular app used a function for physician order entries, specifically for nutrition plan entries [86].

M-Health

Most included studies contained *m-health* components (Fig. 3). Among the various m-health interventions analysed, helplines represented the smallest category. Specifically, four interventions provided emergency telephone support, and one included useful helpline numbers in their app [35, 60, 81, 89, 90].

The predominant categories within the *m-health* domain were clinical support and remote data collection. Nine studies reported interventions with synchronous consultations and data collection [40, 51, 57, 62, 65, 73, 75, 81, 85]. To illustrate, Christodoulou et al. conducted telephone-based cognitive-behavioural screening in ALS patients [85], demonstrating how telemedicine can seamlessly combine remote data collection processes with distance consultations. Another example was the remote application of the ALS Functioning Rating Scale during teleconsultations [62]. An alternative approach identified involving clinical support and remote data collection occurring asynchronously, utilising specially designed devices or mobile applications for data collection [35, 66, 68–70, 88, 90]. In this approach, clinical consultation was offered either on demand or automatically triggered based on the collected data.

Fourteen studies used clinical support without remote data collection, including home exercise programs [40, 82–84], psychological interventions [39, 87] and pure teleconsultation [52, 54, 55, 60, 77–79]. In contrast, 12 studies focussed on pure remote data collection without clinical support. This included, accelerometers [74, 80], physical assessments [63, 64, 67, 76] or the assessment of the patient's nutritional status [86] or disease-related health [37, 46, 89]. Additionally, Cesareo et al. as well as Wasilewska et al. examined remote pulmonary monitoring devices [41, 49].

Barriers and facilitators for the implementation of telemedicine

CFIR was used to assess factors that may facilitate or hinder the implementation of telemedicine. This framework consists of five domains: the inner setting, the outer setting, the implementation process, the intervention characteristics, and the characteristics of the individuals. Relevant information was found in 22 studies, with a predominant focus on patient and carer perspectives [34, 36, 37, 41, 42, 48–52, 55, 62, 63, 66, 69, 73–78, 83]. As a result, no information regarding the inner/outer setting or the implementation process was gathered. All statements focused on the intervention characteristics or the characteristics of the individuals. Thus, the following section is structured according to the two domains and their constructs.

Intervention characteristics

A summary of mentioned barriers and facilitators can be seen in Table 2.

General characteristics This category summarises all barriers and facilitators directly linked to the intervention that could not be categorised elsewhere. The most common barrier encountered during the implementation of telemedicine interventions were malfunctions related to internet connectivity or end devices. Examples included software errors [51], faulty data transmission [34] or a poor internet connection [83]. Additionally, it was reported, that the internet and necessary end devices, such as smartphones, tablets, or computers, were often not available [48, 50, 63].

Relative advantage A major factor for patients was the reduced time and travel burden [34, 51, 52, 62, 76, 78]. In more advanced stages of the diseases travelling with

Table 2 Barriers and facilitators—intervention characteristics according to CFIR framework (Source: own depiction)

General characteristics	
Barrier	• Technical errors • No access to internet/computer/smartphone
Relative advantage	
Facilitator	• Timeliness of data transmission and clinicians' actions • Time effective • Less burdensome/fatigue • Reduced travel time and distance • More comfortable at home/advantage of real-life setting • Telemedicine as only option to receive care • Improved monitoring/communication/care coordination • Not feeling abandoned (during pandemic) • Disability prevents patients from in-person appointments • Clinics unequipped for patients' disability • Appointments more effective due to remotely collected data[a] • Patients/Caregivers more honest via telemonitoring device[a]
Barrier	• Less informal personal contact • Lack of privacy • Lack of physical contact • Feeling trapped in the house • Less personal
Adaptability	
Facilitator	• Adaptability to own schedule • Individualization of mode of communication
Complexity	
Facilitator	• Low burden of intervention • Clear understanding of difficulty/complexity • Number of sessions • Breaks during session • Daily reminders
Barrier	• Long sessions
Design Quality & Packaging	
Facilitator	• Easy to fixate and wear • Accessible packaging • Aesthetical pleasing • Pictures in instructions
Barrier	• Small keys/Insensitive touch panel • Not bite-proof (for children) • Noise
Cost	
Facilitator	• Potentially cost saving
Barrier	• Technology too expensive

[a] Reported by healthcare workers

medical equipment became almost impossible, making telemedicine vital for house-bound patients [78].

Telemonitoring and the remote data collection provided multiple advantages, with patients and caregivers highlighting the timeliness of actions in case of alerts [34, 73]. Continuous monitoring also proved beneficial for in-person visits, as medical staff stated that appointments could be used more efficiently with data being analysed beforehand [69]. Some disadvantages regarding telemedicine were acknowledged. Caregivers and physicians noted the lack of physical evaluation as problematic [51, 52]. Additionally, an emotional distance and a lack of informal encounters between patients and healthcare workers was reported [52, 55].

Adaptability Patients appreciated the flexibility of online exercise programs, which were easier to integrate into their daily routines [83]. It was seen as important to be able to choose the main form of communication [55, 62]. For example, patients with speech difficulties communicating via E-Mail was preferred.

Complexity Interventions were easier implemented if participants were thoroughly informed about the telemedicine service and if a computer-literate person was on-site [78]. The duration and frequency of sessions was another major point. Overall, more frequent, and shorter sessions were perceived as less fatiguing [78].

Design and quality Critical considerations included the presentation, design, and quality of telemedicine products, emphasizing features like accessible closing mechanisms for wearable devices and age-appropriate designs [49, 74].

Cost From a patient's perspective telemedicine was cost-saving due to reduced travel [34, 48]. Nevertheless, acquisition costs could be a barrier for some. Institutional perspectives indicated potential savings, ranging from 20 to 89%, depending on the approach, making costs a crucial factor [50, 77].

Characteristics of individuals

The second domain related to the characteristics of individuals. This includes all stakeholders such as patients, caregivers, and healthcare workers. Table 3 depicts the barriers and facilitators relating to the characteristics of individuals.

Knowledge and Beliefs about the Intervention The CFIR highlights the importance of an individual's pre-existing

Table 3 Barriers and facilitators—characteristics of individuals according to CFIR framework (Source: own depiction)

Knowledge & Beliefs about the intervention	
Facilitator	• Trust in remote monitoring
Barrier	• Lack of information
Self-efficacy	
Facilitator	• Easy to use technology • Confidence in own abilities to use technology
Barrier	• Technophobia
Other personal attributes	
Facilitator	• Younger age • Higher education • Patient empowerment • Existing personal connection between provider and patient
Barrier	• Self-monitoring as confronting

knowledge and beliefs about the intervention [30]. Trust in the intervention was vital for patients using telemonitoring [34, 36, 52, 69, 78]. This includes being confident that the transmitted data was monitored and that providers would act in the case of abnormalities.

Self-efficacy Easy to use technology was seen as an enabler for telemedicine implementation, as it reassured the user in their abilities. Accordingly, barriers arose if patients could not or did not feel confident in using technological devices [50, 51, 69]. Lack of confidence led patients to use technology on rare occasions and only if deemed necessary [36].

Other personal attributes Lastly, this category summarises all personal traits of stakeholders that might impact the implementation of the intervention [30]. Younger, higher-educated patients embraced technology more readily [42, 75]. Another enabler was telemonitoring improving patient empowerment, symptom awareness, and communication [34, 36, 51, 69]. However, some found constant disease confrontation challenging [69]. Lastly, a personal connection with medical staff enabled telemedicine use [36].

Discussion

This systematic review presents a comprehensive overview of the current status of telemedicine applications for patients with NMDs. The primary objective was to classify the identified interventions according to the dimensions of telemedicine. While some studies within this review explored the epidemiology of NMDs, and two interventions provided education for clinical staff, it's clear that certain aspects of telemedicine in public health remain under-studied.

E-health, encompassing health information, an electronic health record or physician order entries/treatment instructions, was comparatively underutilised, with only a subset of interventions included. Moreover, decision support systems were rarely investigated. The predominant focus of most interventions was on clinical support and remote data collection.

The second phase of the analysis concentrated on the implementation process, with a specific focus on identifying barriers and facilitators associated with both the intervention itself and the individuals involved. In comparison to traditional care, telemedicine often demonstrated a relative advantage. The high motivation demonstrated by NMD patients and their caregivers in integrating telemedicine into their care plan is a testament to the potential of telemedicine as a transformative force in healthcare.

Telemedicine was often perceived as a resource-saving, less fatiguing alternative, particularly offering increased accessibility for homebound patients. The lack of physical touch and reduced personal connections emerged as significant barriers. Additionally, the accessibility of technology played a pivotal role, as inadequate design hindered some patients from using telemedicine services. The acceptance and uptake of telemedicine services often depended on the readiness of patients and their caregivers to embrace and adapt to new digital solutions. Recognising the importance of patient empowerment, fostering the development of essential skills and confidence in utilising technology is crucial for enabling patients to actively engage in their healthcare.

Clinical and policy implications

The COVID-19 pandemic created an unprecedented opportunity for the development and implementation of telehealth. Disruptions in healthcare access, caused by social distancing and hygiene guidelines, led healthcare practitioners to expand telemedicine services to ensure the continuity of care [91, 92]. This trend extended to the field of neuromuscular disease care as well [48, 62, 91, 93, 94]. The American Academy of Neurology's "Telehealth Position Statement" endorsed telemedicine, citing benefits such as improved access, reduced costs, and enhanced comfort, aligning with findings in this review [95].

Our findings further highlighted important considerations for the successful implementation of telemedicine. Firstly, it is essential to recognise that not all geographic locations are equally suited for telehealth. Remote areas with insufficient internet or cell phone coverage, as well as low-income households with a lack of digital

technologies, may encounter difficulties in participating in telemedicine interventions [96]. Secondly, careful selection of the target population is vital, as the attitude and willingness of users significantly impact technology uptake [34, 36, 52, 69, 78]. The acceptance and efficacy of telemedicine interventions are inherently intertwined with diverse cultural attitudes towards healthcare and technology.

Therefore, understanding cultural factors is critical to discern how these variables may influence the successful integration of telehealth programs across diverse patient populations. A systematic analysis of cultural competence would provide valuable insights to refine and customise approaches, meeting the distinctive needs of diverse communities. Such considerations not only enhance the inclusivity of telemedicine but also contribute to its overall effectiveness and acceptance among a broad spectrum of individuals.

As the results have shown, it is vital to adapt telemedicine to the specific and evolving needs of patients with NMDs. These needs not only vary from patient to patient but also change over time as the disease progresses [5]. Therefore, when designing telemedicine technology for patients with NMDs, emphasis should be placed on adaptability, flexibility and accessibility [49, 55, 62, 74, 83].

Designing telemedicine technology that caters for the unique challenges faced by patients with physical disabilities and cognitive impairments is crucial for fostering inclusive healthcare [49, 74]. User interfaces need to incorporate accessibility features, such as voice commands, large fonts, and intuitive navigation, to accommodate individuals with motor challenges or cognitive limitations. Additionally, instructions and information must be presented in various accessible formats, accommodating diverse learning needs [97].

Prioritising plain language and ensuring readability at lower literacy levels is essential. This approach not only makes instructions universally accessible but also empowers all patients to effectively participate in telemedicine interactions. By incorporating these considerations into the design, telemedicine can better serve the needs of patients with NMDs, promoting inclusivity and enhancing the overall effectiveness of healthcare delivery [97].

Health policies and regulatory frameworks play a significant role in influencing the development and adoption of telehealth practices. A nuanced understanding of these regulations, encompassing aspects such as licensure, reimbursement, and liability, is essential for gaining comprehensive insights into the complex landscape that shapes and governs telemedicine [96]. The intricate web of reimbursement policies directly influences the economic viability of telemedicine services, impacting both healthcare providers and patients. By navigating and understanding these policy and regulatory intricacies, stakeholders in the telemedicine ecosystem can strategically address and potentially overcome barriers, facilitating a more widespread and effective implementation of telehealth services [96].

This review reveals that telemedicine interventions for patients with NMDs exist but have yet to realise their full protentional. Firstly, the heavy focus on ALS care should be expanded to encompass all diagnostic groups within the NMD spectrum. Especially the high availability of mHealth applications, which could be seamlessly integrated into care plans. This integration has the potential to enhance continuity of care, simultaneously easing the burden on the healthcare system and reducing appointment frequency for patients [69].

The incorporation of long-term patient data through remote monitoring holds numerous advantages [98, 99]. Continuous data collection could offer enhanced insights into disease progression, thereby improving disease management. Given the degenerative nature of most NMDs, there is a speculation that long-term data could help in detecting early signs of deterioration, facilitating quicker adaption of treatments. Furthermore, detailed information about disease progression could contribute to health prognosis, empowering both patients and healthcare professionals to better plan and coordinate care [98, 99]. It is evident that the full benefits of telemonitoring remain undiscovered, making it an important and interesting area for future research. The exploration of these untapped potentials could significantly advance the effectiveness and scope of telemedicine in the context of NMDs.

Research and evaluation opportunities

The current telemedicine landscape yields promising results, particularly in its role in supporting rare disease research through the establishment of disease registries. These registries systematically collect patient data related to disease progression and treatment, forming the foundation for observational studies [100, 101]. These studies offer critical insights into the management and progression of rare disease, contributing to evidence-based clinical decisions and facilitating the recruitment of participants for clinical trial.

National and international patient registries are pivotal for studying prevalence and incidence, enhancing our understanding of rare diseases like neuromuscular disorders [100, 101]. The establishment of global patient registries becomes especially important for pooling data on rare diseases. International collaborations can help bridge the gap in research for understudied NMDs. By

fostering collaboration and sharing data on a global scale, telemedicine-supported registries contribute significantly to advancing our understanding and management of rare diseases.

The results of our systematic review highlight a gap in the research on telemedicine for NMDs. Except for ALS, most NMDs are underrepresented in the current body of literature. Future research should include a more diverse range of diagnostic groups and undertake a comparative analysis of challenges and solutions. This would lead to a higher external validity and faster adaption of telemedicine solutions.

While teleconsultation and remote monitoring for NMDs are well described, other critical domains within telemedicine have received comparatively limited attention. These research gaps should be addressed in the future. Most importantly, implementation science has a critical role in the successful deployment of telemedicine interventions for NMDs. As seen in this systematic review studies, the focus needs to be on patients, caregivers, and health care practitioners, as well as the intervention itself.

It is noteworthy that there is underreporting of crucial aspects, such as the inner and outer settings, as well as the implementation process, in telemedicine interventions for NMDs. Additionally, there is need for research examining the impact of health policies and clinical guidelines on the adoption and implementation of telemedicine. The lack of implementation research has been described in the systematic review by Helleman et al., who analysed telemedicine for ALS patients [25]. Implementation science is needed to improve the efficiency and uptake of future telemedicine interventions for NMDs [102].

While our systematic review focused on highlighting the barriers and facilitators of telemedicine, we fully recognise the importance of addressing the validation challenges associated with digital health data. Future research and healthcare policies should emphasise the need for robust validation processes to ensure the reliability and clinical relevance of digital outcomes in telemedicine interventions.

Limitations

Despite an extensive search string, additional search terms might have yielded more results, especially considering synonyms for neuromuscular diseases. A more specific search for individual diagnostic groups would have been more inclusive, but the sheer number of NMDs made this unfeasible.

The literature databases used represent common sources of clinical evidence, but they may not comprehensively cover health policies, management, and health education related to NMDs, which might be found in other types of databases.

The absence of experimental study designs in the individual studies was notable, with most included studies being cross-sectional or observational. However, as this review aims to provide an overview of interventions, this description suffices.

The majority of included studies are from high-income countries, and the extent of telemedicine utilisation in low- and middle-income countries remains unclear. The variation in target population size and time horizon in NMD research reflects the complexity and rarity of these conditions, suggesting a need for longer follow-up times in future studies to better describe long-term outcomes.

Conclusion

This systematic review offers a comprehensive view of the telemedicine landscape in the context of NMDs. While domains like teleconsultation and telemonitoring have received extensive attention and reporting in the literature, other critical domains, such as decision support tools and informational support, are notably lacking in research and documentation. To further understand, develop and implement telemedicine solutions and to close existing gaps in NMD-specific healthcare provision, policies and guidelines are needed. By actively integrating telemedicine into existing healthcare plans and maintaining a commitment to ongoing updates and improvements, healthcare systems can optimise care delivery, enhance patient outcomes, and ensure that individuals with NMDs receive the high-quality care they deserve. In addition, more high-quality studies are needed to close research gaps concerning the implementation process of telemedicine and prove the respective efficiency and effectiveness in the long run.

Abbreviations

ALS	Amyotrophic lateral sclerosis
ASS	Anti-synthetase syndrome
BMD	Becker muscular dystrophy
CFIR	Consolidated framework for implementation research
CM	Congenital myopathy
CMD	Congenital muscular dystrophy
CMT	Charcot-Marie-tooth disease
DM	Dermatomyositis
DMD	Duchenne muscular dystrophy
EDMD	Emery-Dreifuss muscular dystrophy
FSHD	Facioscapulohumeral muscular dystrophy
HSP	Hereditary spastic paraparesis
ICD	International classification of diseases
JDM	Juvenile dermatomyositis
LEMS	Lambert-Eaton-myasthenic-syndrome
LGMD	Limb-Girdle muscular dystrophy
MyD	Myotonic dystrophy
MD	Muscular dystrophy
MDA	Muscular dystrophy association
MFM	Myofibrillar myopathies
MG	Myasthenia gravis

MGSD	Muscle glycogenosis
MP	Myopathy
NM	Necrotizing myositis
NMD	Neuromuscular disease
OM	Overlap myositis
PD	Pompe disease
PM	Polymyositis
PPS	Post-Polio syndrome
RCT	Randomized controlled trial
SBMA	Spinal and bulbar muscular atrophy
SMA	Spinal muscular atrophy
TTR-FAP	Transthyretin familial amyloid polyneuropathy

Supplementary Information

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Supplementary Material 1.

Supplementary Material 2.

Supplementary Material 3.

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DS and KS prepared the study protocol as well as performed the literature search and study selection. DS conducted the data extraction and analysis. The report was written by DS with contributions by KS. JB and KN supervised the complete process. All authors read and approved the final manuscript.

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Author details
[1]Chair of Healthcare Management and Health Services Research, University of Bayreuth, Bayreuth, Germany. [2]Chair of Digital Health and Innovation, Department of Public Health, School of Psychology and Public Health, La Trobe University, Melbourne, Australia.

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