Occupational therapy, physical therapy and speech therapy for persons with neuromuscular diseases

An evidence based orientation

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Contents

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Chapter 1
General introduction

Introduction

Persons with a chronic neuromuscular disease (NMD) may experience a host of problems in daily occupations and may be more or less severely restricted in activities and participation in life roles. They continuously have to deal with changes in their health condition as a result of the chronic progressive nature of the disease. Since there is no cure for most NMDs, it is important to provide support and care on a regular basis, both to the persons with NMD and their families, helping them to cope with the disease and continue to participate in meaningful roles[1]. The aim of the current thesis is to develop a better understanding of how allied health professionals, specifically occupational therapists (OT), physical therapists (PT) and speech therapists (ST) can help to achieve these objectives.

There is wide variety in referral, content and duration of OT, PT and ST for persons with NMD. Referral largely depends on the knowledge and experience of the referring physician with OT, PT and ST. Most frequently, persons are referred to PT in primary care. The content and duration of PT vary and not only depend on the type and severity of the impairments in body functions and restrictions in activities and participation, but also on the expertise of the PT. It is not unusual for persons with a NMD to have PT continuously for many years, the costs of which are fully reimbursed in the Netherlands. For OT and ST, the situation is different. In general, there is limited knowledge among referring physicians, among persons with NMD and even among professionals themselves regarding the treatment options of OT and ST for persons with NMD.

In current thesis we aim to increase this knowledge for all involved. A way to gain knowledge, experience and new insights is by regularly seeing persons with a NMD and by explicit and conscious professional reasoning, often referred to as clinical reasoning. Within this reasoning process, the professional integrates knowledge regarding the client situation, the therapy context, theory and research, and personal beliefs and ideas about therapy outcomes[2].

The premise that service provision of OT, PT and ST for persons with NMD has been inefficient was the starting point of our thesis. This premise led to a comparative cohort study in which current allied health care utilization was compared with allied health care advice from a neuromuscular expert centre. This advice was based on single OT, PT and ST consultations and multidisciplinary meetings. In these consultations client expertise, professional expertise and scientific evidence were combined, as in

evidence based practice (EBP). These elements of EBP are key elements in current thesis. Following a brief introduction on NMD, the elements of EBP are further elaborated on.

Neuromuscular diseases

Neuromuscular diseases (NMD) can be classified anatomically, making clear in which part of the peripheral nervous system the consequences of the disease are present. Four categories are distinguished[3]: 1) Motor neuron disorders such as amyotrophic lateral sclerosis (ALS) and spinal muscular atrophy (SMA), which may involve motor neurons in the brain, spinal cord, and periphery, and ultimately weaken the muscles; 2) Motor nerve root disorders and peripheral neuropathies such as Charcot-Marie-Tooth disease (CMT) and Guillain Barré Syndrome, that not only affect motor but also sensory nerves; 3) Neuromuscular transmission disorders in which the neuromuscular junction may also be directly involved in diseases such as myasthenia gravis; and 4) Muscle disorders such as Duchenne and Becker muscular dystrophies, myotonic muscular dystrophy (MD), facioscapulohumeral muscular dystrophy (FSHD), inflammatory myopathies or limb-girdle muscular dystrophy (LGD). Most of these conditions are slowly progressive and involve several impairments in functions such as muscle weakness, sensory loss, pain, fatigue and autonomic dysfunction in varying combinations. Some types of neuromuscular diseases, like myotonic dystrophy, are in fact multisystem disorders with involvement of many organs, including impairments in the muscular, respiratory, cardiac, central nervous, endocrine and ocular functions and structures[4]. The impairments in various functions and structures may result in a variety of limitations in activities and restrictions in participation.

In the Netherlands there are about 100.000 persons living with an inherited or acquired NMD, sometimes for a restricted period of time, but often throughout their whole life[5]. There are over 600 different NMD with a wide diversity of symptoms, prevalence, hereditary, aetiology, progression and treatment[6]. The majority is rare and these are mainly the hereditary NMD. A survey of the world literature on population frequencies of various inherited NMD affecting children and adults resulted in 1991 in a conservative estimate of 1 in 3500 of the population to have a disabling inherited NMD[7]. Most likely this prevalence has increased due to the increased longevity of persons with NMDs. Myotonic dystrophy and hereditary motor and sensory neuropathy (HMSN), also known as Charcot-

Marie-Tooth (CMT) disease are most common with prevalence's of respectively 1/20.000 and 1/1.000[6].

Evidence based practice

Since the introduction of evidence-based medicine, it has evolved to evidence based practice (EBP), emphasizing that it is not only applicable to medical professions but also to many other health care services[8]. The most widely used definition is from Sackett et al. (2000) who describe EBP as 'the conscientious, explicit and judicious use of current best evidence in making decisions about the care of individual patients[8;9]. Most definitions of EBP distinguish three elements: 1) professional expertise, defined as the proficiency and judgment that individual clinicians acquire through clinical experience and clinical practice; 2) best available external scientific evidence from systematic research; 3) predicaments, rights, preferences, norms and values of individual clients[8-11]. Although persons with chronic illnesses become experts in managing their disease, their knowledge and experience have long been an untapped resource[12]. There are situations in which the clients know more about their diseases than the professionals[13]. It is therefore justified to distinguish both, client expertise as well as professional expertise. Also, for health care professionals it is important that they are aware of their own beliefs and values when providing services as these influence the way support is provided[14]. Expertise of clients and professionals thus both comprise knowledge, expertise, norms and values. As all these elements are context-specific, the context is also an important element in EBP[8]. The next paragraphs will describe aspects related to client expertise, professional expertise, scientific evidence and the context, which includes the practice context for OT, PT and ST for persons with NMD as well as the societal context (Figure 1).

Client expertise

In a practice context, many health care providers focus on diseases and their management, rather than on people, their lives and their health issues [15]. For evidence based decision making in OT, PT and ST, information on the disease and its course is important, but using client expertise involves much more than that. Each client attending OT, PT and/or ST has unique physical, psychological, emotional, environmental and personal perspectives, knowledge and experience influencing their activities and participation. Gaining insight and understanding of these perspectives can be obtained by

using observations and tests and, above all, by using a narrative approach asking persons to tell about their lives. Client-centered practice and qualitative research both explicitly make use of client expertise and aim to increase understanding of the client perspective.

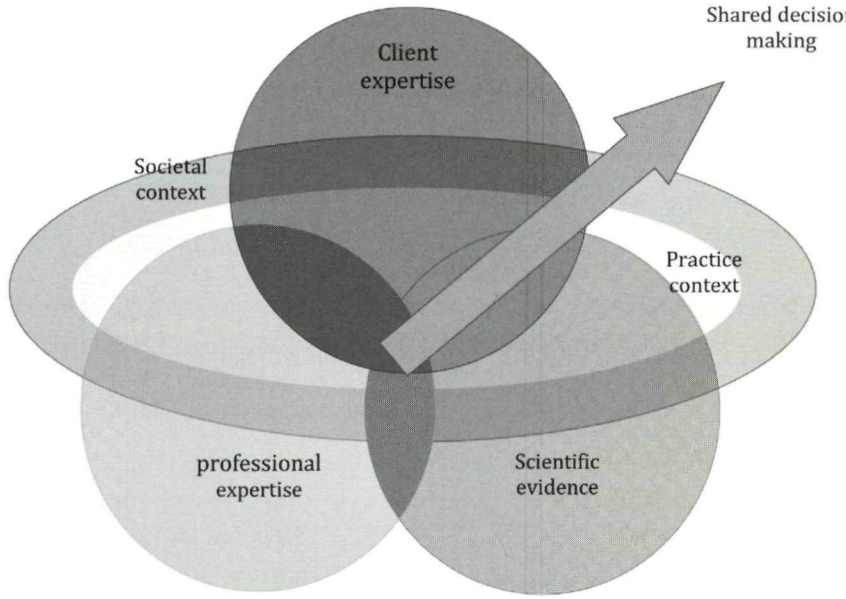

Figure 1. Elements of evidence based practice (EBP) in a practice context and societal context.

Client-centered practice

A range of definitions and related concepts can be found to describe client-centered practice[14-16]. Also many terms have been used to describe concepts centered on people being partners in health care. These terms include client-centered, patient-centered, patient-focused or family-centered[14]. An early definition described patient-centered medicine as an approach whereby the health care provider uses the patient's knowledge and expertise to guide the interaction within the consultation[15]. This shows how evidence based practice and client-centered practice are congruent with and complementary to each other for shared decision making[17]. A later definition described the patient-centered approach as one whereby the health care providers try to enter the patient's world, to see the illness through the patient's eyes[15;16]. Six

interacting components of the patient-centered process have been described: 1) exploring both the disease and the illness experience; 2) understanding the whole person; 3) finding common ground regarding management; 4) incorporating prevention and health promotion; 5) enhancing the patient-professional relationship and 6) establishing realistic goals[16]. The various models stress the important contributions that both the client and the practitioner bring to the intervention and the strength of combining these[16]. Other components of being patient-centered include taking the patient and his or her problems, ideas and expectations seriously, involving the patient in decisions, giving information to enable patients to take responsibility for their own health, and feeling responsible for non-medical aspects of the presented problem[16;18].

In a review on interventions to promote a patient-centered approach in clinical consultations, two main features were identified as part of a broad definition of patient-centered care[15]: 1) healthcare providers share control of consultations, decisions about interventions or the management of the health problems with patients; and/or 2) health care providers focus on the patient as a person, rather than solely on the disease, in consultations. Within this definition, shared treatment decision making is an indicator of patient-centered care[15]. The Dutch advisory board for health care (Raad voor Volksgezondheid en Zorg [RvZ]) recommends an even more central role for the client in their health care management[13]. In the advice report Health 2.0, the client participates in communities that support him/her to stay or become healthy or to manage their chronic illness. In Health 2.0 clients are seen as persons and partners in care and they have autonomy regarding their lives and health care interventions[13].

Client perspective from qualitative studies

Several qualitative studies have investigated the illness experience of persons with a NMD and how they have dealt with it[1;19-24]. Most obvious were restrictions in mobility and increased fatigue and feebleness. The persons described psychosocial consequences of muscular dystrophy as well as stigma when the disability had become more obvious. In spite of reported distress, several persons experienced better psychological adaptation over time[20]. All studies stressed the importance to gain insight in the illness experience of persons with a NMD in order to offer the best possible support[1;19-24].

One study aimed to elucidate the next of kin's experiences when an adult member has muscular dystrophy. This study revealed the need for health care professionals to understand the next of kin narrated meaning of changes when an family member has a progressive disease[22]. One other study described conceptions and experiences of the hereditary aspect of muscular dystrophy from both the patients' and the next of kin's' perspective. It was found that heredity has a prominent place in the thoughts and feelings of the family and that families with a muscular dystrophy need medical information and the opportunity for genetic testing as well as support and counseling in coming to terms with living with a hereditary disease[21].

Professional expertise

Professional expertise is based on knowledge and experience, norms and values and is influenced by the professional domain[2]. A profession's domain articulates its sphere of knowledge, societal contribution, and intellectual or scientific activity. Defining the knowledge and skills needed for optimal allied health care services for persons with NMD appears difficult and therapists often lack expertise in the treatment of persons with NMD[25;26]. In this paragraph the professional domains of OT, PT and ST are described and an attempt is made to describe specific skills and experience required for the assessment and management of persons with NMD. Finally, the professional expertise in EBP or professional reasoning is explained.

Occupational therapy

The OT profession's domain centers on enabling occupation[27]. The World Federation of Occupational Therapists[28] has defined OT as a client-centered health profession concerned with promoting health and well being through occupation. The primary goal of OT is to enable people to participate in activities of everyday and societal life. OTs achieve this outcome by working with people and communities to enhance their ability to engage in the occupations they want to, need to, or are expected to do, or by modifying the occupation or the environment to better support their occupational engagement. OTs believe that participation can be supported or restricted by the physical, affective or cognitive abilities of the individual, the characteristics of the occupation, or the physical, social, cultural, attitudinal and legislative environments. Therefore, OT practice is focused on enabling individuals to change aspects of their person, the occupation, the

environment, or some combination of these to enhance occupational participation[28].

Hill and Phillips have described the specific experience required for OT in NMD as experience with long/term chronic conditions and knowledge of their particular needs with reference to education, work and education, living, family life, and social and leisure activities[26]. In current thesis, the OT assessment and interventions for NMD are described as follows: The OT assessment consists of a client-centered interview in which the client is asked to describe an ordinary day and highlight the problems encountered in occupational performance. In addition, the OT observes the performance of a daily task looking at safety, efficiency aspects and discusses these with the client. The following OT interventions for persons with NMD have been distinguished in this thesis: training of activities of daily living (including self care, mobility, work, household tasks, leisure) or training of skills, including (fine) motor skills, advice and instruction in the use of assistive devices, provision of splints and slings, counseling on energy conservation strategies, educating patient, family and caregivers or a combination of the above.

Physical therapy

The definition of PT according to the World Confederation for Physical Therapy (WCPT)[29]: PT provides services to individuals and populations to develop, maintain and restore maximum movement and functional ability throughout the lifespan. This includes providing services in circumstances where movement and function are threatened by ageing, injury, disease or environmental factors. Functional movement is central to what it means to be healthy. PT is concerned with identifying and maximizing quality of life and movement potential within the spheres of promotion, prevention, treatment/intervention, habilitation and rehabilitation. This encompasses physical, psychological, emotional, and social well being. PT involves the interaction between physical therapist, patients/clients, other health professionals, families, care givers, and communities in a process where movement potential is assessed and goals are agreed upon, using knowledge and skills unique to PTs[29].

Hill and Phillips have described the specific expertise of PT for people with muscle disease as requiring skills in both neurological and musculoskeletal PT, experience in treating muscle conditions and the confidence to treat patients with rare disorders[26]. In current thesis, the PT assessment and interventions for NMD are described as follows: The PT

assessment focuses on muscle strength, mobility and balance and its relation to activities like walking, transferring and lifting and carrying. In a semi-structured interview problems in performing these activities are elicited, as well as any prior attempts to deal with them. Based on the problems expressed, a set of functional tests for balance and walking and functional observations like performing transfers and stair climbing are recorded. At the level of impairment, an overview of muscle strength, muscle length, joint mobility and stability, pain and tenderness is obtained. The following PT modalities for persons with NMD have been distinguished: exercise therapy such as muscle strengthening exercises, mobilizing exercises, aerobic exercises, breathing exercises; and other interventions which could be training of skills such as transfers or taking stairs or a combination of these interventions.

Speech and language therapy

The domain of ST includes human communication behaviors and disorders as well as swallowing. The overall objective of ST services is to optimize individuals' ability to communicate and/or swallow in natural environments, and thus improve their quality of life. This objective is best achieved through the provision of integrated services in meaningful life contexts[30]. A ST's occupation entails the development, improvement and recovery of four vital aspects of human communication: speech, the human voice, language and hearing, including the primary functions at the basis of these aspects, i.e. swallowing[31]. The ST is the professional responsible for the prevention, assessment, treatment and scientific study of human communication and related disorders. In this context human communication encompasses all those processes associated with the comprehension and production of oral and written language, as well as appropriate forms of non-verbal communication[32].

According to Hill and Phillips, STs in muscle diseases need skills and expertise in the assessment and management of dysphagia[26]. In current thesis, the assessment and interventions for ST in NMD are described as follows: The assessment focuses on swallowing and communication. Following an interview, a standardized assessment of oral motor functioning and speech is conducted. Also swallowing tests are used to obtain information about swallowing abilities. In this thesis we distinguished various ST interventions for persons with NMD: information and advice or teaching compensatory strategies. Information and advice incorporated

dietary modification, augmentative and alternative communication or instruction of the patient and relatives. Compensatory strategies included teaching swallowing maneuvers and/or strategies to improve intelligibility.

Professional reasoning and shared decision-making

Professional reasoning about decisions regarding the care for clients is about how therapists put science and art of practice together[2]. During professional reasoning, etiology of diseases, insight in the client situation, diagnostic and therapy options, theory and research are integrated with professional expertise. The professional expertise, acquired through practical experience, is used in the various stages of the therapy process[9;33;34]. The professional uses interview skills, assessments and theoretical knowledge to gather information on the client perspective. This involves several ways of professional reasoning including narrative reasoning to understand the life history of the client and the meaning of the illness for the client and their aims for the future[2]. This is combined with scientific reasoning in which the professional uses knowledge and experience regarding the diagnosis, prognosis, course of the disease and current health status with the desired situation regarding abilities and restrictions in activities and participation. For scientific reasoning the available scientific evidence regarding the assessments and interventions is used. The problems identified are translated into research questions, preferably from a client's perspective[33]. After framing the question, research evidence is searched and appraised or professional practice guidelines examined and the applicability of the results to a particular client determined. Pragmatic reasoning is also applied to find out what is feasible in the context[2]. And finally, in the care for NMD, aspects of ethical reasoning may arise when desires of persons are in conflict with their physical capabilities and prognosis. This all leads to the collaborative process with the client in shared decision making. The professional expertise has influence on the quality of both the discussion of client and research evidence, and the shared decision making. Finally, professional expertise and client expertise are used to evaluate the effect of therapy, which may result in new goals and researchable questions and the therapist's and client's accumulation of expertise, which has the potential to affect future decision making[33].

Professionals have acknowledged that they lack expertise in research retrieval and analysis, which is a barrier to the integration of research evidence into practice[35]. A helpful tool to bridge the gap between scientific

evidence and clinical practice is the development of evidence based and consensus based guidelines. Evidence based guidelines regarding OT, PT and ST for persons with NMD are lacking and it is one of the future challenges for professionals and clients to develop such guidelines.

Scientific evidence

Ideally, each decision regarding the professional analysis of the problem, testing and choice of intervention is based on the best available scientific evidence[36]. This requires formulation of the right questions and translating these into searchable questions, often with the use of the PICO method (P: Patient, Population or Problem; I: Intervention or exposure; C; Comparison and O: Outcome) followed by a search in the appropriate databases. In current thesis, literature searches have been carried out for the population with a NMD (P), for OT, PT and ST interventions (I and C) and outcome measures at the level of body functions, activities and participation or quality of life (O).

Initially, the Cochrane database was searched for systematic reviews of randomized clinical trials (RCTs) and controlled clinical trials (CCTs), as these are considered the golden standard in EBP. However, no Cochrane reviews existed regarding OT for persons with NMD. Three Cochrane reviews investigated specific PT interventions for persons with specific types of NMD and all three concluded that there was insufficient evidence to establish the benefit of exercise therapy[37-39]. For ST, one Cochrane review on interventions for swallowing difficulties (dysphagia) in persons with chronic muscle disease also concluded that there is insufficient evidence[40].

The majority of NMD are rare, which makes it difficult to carry out RCTs and CCTs with large enough groups of participants. Observational designs with a long term follow-up might be more suitable. Therefore, the systematic reviews carried out in current thesis, also included studies other than RCTs and CCTs.

The applicability of scientific evidence depends on factors related to the client, the professional as well as the context[34]. To know whether an intervention might be effective for an individual, it is important that the client matches the in- and exclusion criteria defined in the study. Most studies have strict in- and exclusion criteria, which may be the reason for professionals to dismiss findings from an RCT when their clients are different from those in a trial[41]. For the professional, it is important to know which skills or

knowledge is needed for assessment and management of persons with NMD and whether additional education or training is necessary. There may be barriers such as the availability of therapeutic material, finances, health care systems, opinion or culture within a (professional) organization[34]. In a research context, standardized treatment protocols are often required with extensive quality control to decrease errors in treatment delivery. Hence, a research context may not reflect the clinical practice environment[41]. Moreover, the clinical practice environment does not reflect the context in which persons live and were they have to cope with the problems as a result of their chronic condition.

Context

Two contexts are distinguished (Figure 1): practice context and societal context. These contexts are also described in the Canadian Practice Process Framework (CPPF), a framework for evidence-based, client-centered occupational enablement[42]. The practice context is where the client and the professional meet. Both the practice context and societal context have physical, social, cultural and institutional characteristics, such as the models of service delivery and the health care system[42].

Persons with NMD may receive OT, PT and ST in primary care i.e. community or in an expert centre for NMD. Although the community context is most preferable for persons to be treated in, the provision of OT, PT and ST in the community is highly variable and has been described as fragmented, inadequate and even deficient[26;43-45]. In the Netherlands, a general practitioner has a mean number of 15 persons with a NMD in practice [5]. A community practice for OT, PT or ST will have even less persons per year, which means that it is impossible to gain and built expertise regarding the assessment and management of persons with NMD.

Integrated approaches have been advocated incorporating all aspects of NMD including genetic and medical resources as well as rehabilitation and community services for persons and family members, not just at discrete moments but across the continuum[43;45]. Such approach may work well for prevalent diagnoses like Parkinson's disease and multiple sclerosis, but is difficult for NMD, as there are only few centres of expertise in NMD, for which it is less easy to have close links with the many different communities[46].

In the Netherlands there are several diagnostic centers and rehabilitation centers with teams specialized in the treatment for persons with NMD[5]. In these centers integrated medical care and rehabilitation

services are available. As most persons with a NMD receive community services[26;43;45;47], ways need to be found to transfer expertise from specialized teams to professionals in the community.

The Dutch patient support organization (Vereniging Spierziekten Nederland [VSN]) has initiated and supports a network for OTs and PTs with expertise NMD with the aim to increase and share their knowledge and experience[5;48]. The therapists of this network work in the diagnostic centers or specialized rehabilitation centers. Although this network aims to support therapists in community, community therapists are often unaware of this network. Several strategies have been used to increase awareness, such as a website (www.vsn.nl) including a list of expert professionals for each region, regional meetings with community professionals, articles and presentations[48]. A website for health professionals (www.hulpverleners.vsn.nl) is being developed[48]. Participation in professional networking including the use of internet communities are increasingly promoted to exchange and increase professional knowledge and experience[13].

As the government encourages taking own responsibility and increasing self-management skills in persons with chronic diseases and their health care professionals, they have allowed for direct access to OT, ST and PT without medical referral in 2011. This stresses the importance to share knowledge and experience in the field of NMD.

Current studies were carried out from the Neuromuscular Centre of Nijmegen (NMCN) in the Radboud University Nijmegen Medical Centre. In this centre different professionals with expertise in the field of NMD work together in a multidisciplinary team consisting of medical and allied health care professionals, including OT, PT and ST. Most persons visiting the NMCN are from outside the region. Following multidisciplinary assessments, these persons may be referred to OT, PT and ST within the NMCN, a rehabilitation centre or to OT, PT and ST in the community. All persons receive a written report and advice regarding the treatment goals, and suggestions for the volume (duration) of therapy. So far, it was unclear whether this advice was received, appreciated and implemented by persons with NMD and their OTs, PTs and STs in the various contexts.

Aims and outline of this thesis

This thesis is divided into three parts reflecting the elements of evidence based practice as well as the chronological process by which the knowledge and experience regarding OT, PT and ST for persons with NMD was built.

Part I: Professional perspective

Part I was based on the premise that referral to and utilization of PT, OT and ST was not efficient and that a multidisciplinary approach with expert consultations would result in more efficient referral to and utilization of OT, PT and ST for persons with NMD. *Chapter 2* presents a comparative cohort study in which the usual volume of OT, PT and ST is compared with the volume of therapy suggested by OT, PT and ST of the NMCN based on one-off consultations and a multidisciplinary meeting and advice. A cost analyses was carried out to get insight into the costs and possible savings of such multidisciplinary approach. *Chapter 3* explores the extent to which the OT, PT and ST advices were implemented with regard to the volume of therapy suggested. This study also explores whether factors at the level of the advice, the person with NMD, the allied health professional of the context might have influenced implementation of the advice.

Part II: Scientific perspective

Three systematic reviews were conducted to summarize scientific evidence for OT, PT and ST for persons with NMD. These reviews not only included evidence from RCTs and CCTs, but also from uncontrolled pre-post designs. *Chapter 4* presents the results of the systematic review on the efficacy of OT for adults with NMD, *Chapter 5* summarizes the effectiveness of exercise therapy and other types of PT for persons with NMD, and in *Chapter 6* the evidence for ST for persons with NMD is presented.

Part III: Client perspective

Because a single consultation within limited time frame and within a practice context differs from the context where persons live, it does not provide full understanding of the illness experience of persons. Therefore, it was felt that the client perspective was not well understood. This was especially the case for persons with the most prevalent and most complex disease myotonic dystrophy type 1. It was also felt that the perspective of partners should be incorporated because a chronic NMD also affects the lives of partners and also of both of them as a couple. *Chapter 7* provides better insight and

understanding of experiences of couples living with myotonic dystrophy type 1 with a qualitative study using in-depth interviews.

General discussion

Chapter 8 reflects on the findings from the various studies representing the professional perspective, the scientific perspective and client perspective. The perspective regarding service delivery for chronic conditions was added using the Chronic Care Model. This model includes determinants of effective chronic illness care and the nature of practice systems that facilitate good care and good patient outcomes. These elements may assist in improving service delivery of OT, PT and ST in persons with NMD. The discussion concludes with recommendations for future service delivery and research regarding OT, PT and ST in NMD.

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Part I: Professional perspective

Chapter 2
Referral of patients with neuromuscular disease to occupational therapy, physical therapy and speech therapy: usual practice versus multidisciplinary advice

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Abstract

Purpose: To compare the volume of occupational therapy (OT), physical therapy (PT) and speech therapy (ST) as currently received by patients with neuromuscular diseases with the volume of OT, PT and ST recommended by a multidisciplinary team.

Method: The use of OT, PT and ST was studied retrospectively and prospectively in a reference group (n=106) receiving usual care and in an intervention group (n=102) receiving advice based on multidisciplinary assessments. A cost analysis was made and the implementation of the advice was evaluated at 6 months.

Intervention: Multidisciplinary assessments consisted of a single consultation by OT, PT and ST each, followed by a multidisciplinary meeting and integrated advice.

Outcome variables: volume (frequency times duration) of therapy, relative over- and underuse of therapy and costs of therapy and intervention.

Results: Compared to the multidisciplinary advice, there was 40% underuse of OT among patients with neuromuscular disease. For PT, there was 32% overuse and 22% underuse; for ST, there was neither over- nor underuse. Some 40% percent of patients received once-only advice regarding ST compared to 27% regarding OT and 19% regarding PT. The costs of the multidisciplinary advice were estimated at €245 per patient. If fully implemented, our multidisciplinary approach would result in a mean cost savings of €85.20 per patient. The recommended therapy had, however, been implemented only partially at 6 months follow up.

Conclusions: Some patients with a neuromuscular disease do not receive any form of allied health care, whereas they should. Among patients with neuromuscular disease who do receive some form of allied health care, quite a few receive these treatments for too long periods of time. Ways need to be developed to improve implementation of the multidisciplinary advice and to obtain a more favourable balance between its costs and benefits.

Introduction

The prevalence of neuromuscular disease (NMD) in the Netherlands is estimated at 6.25 per 1,000 (100,000 patients in total). Patients with NMD constitute a highly heterogeneous group in terms of incidence, heredity, aetiology, prognosis and functional impairments[1-3]. As a result, it is difficult to identify which patients will benefit from allied health services, such as physiotherapy (PT), occupational therapy (OT) and speech therapy (ST). Patients who suffer from a rapidly progressive NMD such as amyotrophic lateral sclerosis (ALS), need multidisciplinary assessment and treatment[4]. Most adult patients suffer from a mildly progressive NMD, and may require some form of allied health therapy in primary care. Our impression is that in current practice, many patients receive PT but only few patients receive OT or ST. Obviously, referral patterns play a key role in this respect.

To our knowledge, no guidelines or criteria exist for referral of patients with NMD to PT, OT or ST in primary care. As a result, current referral practices seem arbitrary and show considerable variation. The issue is further complicated by the paucity of data on outcome of OT, PT or ST in this heterogeneous group of patients. Regarding PT, one review examined the efficacy and safety of strength training and aerobic exercise training in patients with muscle diseases, based on two randomised trials. Its major conclusion is that there is insufficient evidence of its effectiveness in these patients[5]. There are no controlled clinical trials of OT in patients with NMD. Two uncontrolled trials have been conducted, evaluating the effect of a hand-training programme for patients with myotonic dystrophy type 1[6] and Welander distal myopathy[7]. Both studies showed a positive treatment effect, but the number of patients in these trials was low (5 and 12 respectively), and only a limited part of OT was evaluated. Also with respect to ST in patients with NMD, no evidence exists of its effectiveness[8].

Results from exploratory studies describing experiences and functional abilities in patients with different types of NMD, such as muscular dystrophy[9] or critical illness polyneuropathy[10], may assist in establishing the best possible treatment and support in these patients. From such studies, it may be concluded that a coordinated team approach is essential for achieving optimal outcomes in complicated or complex illness involving multiple extremities and/or multiple organ systems, or an illness that creates disability in the performance of basic activities of daily life (ADL)[10;11].

To ensure that patients with NMD receive appropriate OT, PT or ST, we adopted a multidisciplinary approach, involving expertise from the fields of

neurology, rehabilitation medicine, PT, OT and ST. Apart from the medical diagnosis and prognosis, an assessment was made of the patients' limitations in activities and participation. Upon completion of the assessment, some patients received advice and did not need further treatment. If further treatment was advised, the objectives, content, and expected duration were stated explicitly. In this paper, we describe our experience with such a multidisciplinary assessment of patients with NMD who attended the Neuromuscular Centre Nijmegen. Current utilization of OT, PT and ST (usual care) was documented and compared with the utilisation of OT, PT and ST as recommended by the multidisciplinary team (experimental care). Specifically, the following questions were addressed:

- Did recommendations regarding content and duration of allied healthcare given by the multidisciplinary team differ from actual allied healthcare utilization?
- How frequently was only a single consultation, consisting of assessment and counselling, considered appropriate?
- How did the patients rate the assessment and the recommendations for allied health care?
- Were the recommendations implemented at six months follow up?
- How is the balance between costs and savings of the intervention?

Methods

Study design

A comparative cohort study was carried out with patients attending the Neuromuscular Centre Nijmegen. One cohort served as a reference group and received usual care. A second cohort served as the intervention group and received multidisciplinary assessment and advice (Figure 1). The cohorts were compared at baseline regarding patient characteristics and OT, PT and ST received during the previous six months (Figure 1: comparison 1). In the reference group, OT, PT and ST at baseline and at 6 months follow up were compared to monitor changes over time in case of usual care (Figure 1: comparison 2). In the intervention group, actual OT, PT and ST utilization at baseline were compared with recommendations from the multidisciplinary team (Figure 1: comparison 3). To evaluate changes as a result of the multidisciplinary assessment and advice, OT, PT and ST at baseline and at 6 months follow up were compared (Figure 1: comparison 4). Approval for this study was obtained from the Medical Ethics Committee of the Radboud University Nijmegen Medical Centre.

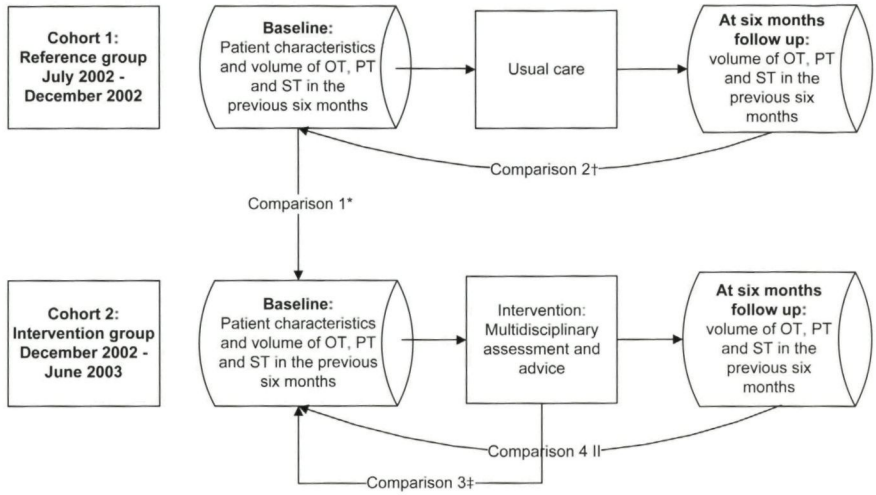

Figure 1. Design of the study

* Comparison 1: Comparison of the two cohorts at baseline in terms of patient characteristics and volume of OT, PT and ST received in the previous six months (amount of OT, PT and ST at baseline). Purpose: To check for comparability of the two cohorts at baseline.

† Comparison 2: Comparison of the volume of OT, PT and ST at baseline with the volume at six months follow up, when usual care is given. Purpose: To check for any changes in the volume of allied care in the absence of a specific intervention.

‡ Comparison 3: Comparison of the amount of OT, PT and ST at baseline with the amount advised by the multidisciplinary team. Purpose: To assess appropriateness of current service provision, by reference to expert opinion.

|| Comparison 4: Comparison of the volume of OT, PT and ST at baseline with the volume at six months follow-up, following the multidisciplinary advice. Purpose: To assess extent of implementation of recommendations.

Patients

Consecutive patients visiting the Neuromuscular Centre Nijmegen from July 2002 to June 2003 were approached for participation. Inclusion criteria were: 1) probable or definite NMD according to the medical records; 2) age 18 years or older; and 3) sufficient command of the Dutch language. Patients of the reference group were recruited from July 2002 until December 2002. Patients of the intervention group were recruited from December 2002 until June 2003.

Intervention: multidisciplinary assessment and advice

The multidisciplinary assessment comprised consultation of the OT, PT and ST (in random order) of the Neuromuscular Centre Nijmegen. The OT assessment focused on problems in the performance of daily activities. The OT conducted a client-centred interview in which the patient was asked to describe an ordinary day and highlight the problems encountered in occupational performance. In addition, the OT observed the patient while performing a daily task (making a hot drink and a sandwich).

The PT assessment focused on muscle strength, mobility and balance and its relation to activities like walking, transferring and lifting and carrying. In a semi-structured interview problems in performing these activities were elicited, as well as any prior attempts to deal with them. Based on the problems expressed, a set of functional tests like a Berg Balance scale[12], 6 minute walking distance[13], analysis of the gait pattern and functional observations like performing transfers and stair climbing were recorded. At the level of impairment, an overview of muscle strength, muscle length, joint mobility, pain and tenderness was obtained. In the analysis the Rehabilitation Problem Solving (RPS) form[14] was used to relate disease, impairments and activities and to decide if PT might be beneficial to the patient.

The ST assessment focused on swallowing and communication. Following an interview, a standardized assessment of oral motor functioning and speech was conducted. Also, two quantitative swallowing tests, the dysphagia limit and the timed test, were used to obtain information about swallowing abilities[15;16].

After the assessment, all therapists registered whether there was an indication for further therapy. If advice was given during the assessment, but there was no need for further treatment, this was registered by the therapist as a single advice. When further treatment was deemed necessary, treatment goals, treatment modality, and expected duration were specified.

The findings and recommendations were summarised and discussed during a multidisciplinary meeting with a neurologist, rehabilitation physician, occupational therapist, physiotherapist, and speech therapist. This resulted in an integrated multidisciplinary advice regarding appropriate OT, PT and ST. This advice was sent to the patient and relevant health care professionals (general practitioner, neurologist, rehabilitation physician and allied health professionals).

Outcome measures

- The 36-item short-form (SF-36)[17] and Euroqol 5D[18] were used to describe functional health status and valuation of individual health state, respectively. The SF-36 is composed of 36 questions and standardized response choices, organized into eight multi-item scales: physical functioning, social functioning, role limitations due to physical problems, role limitations due to emotional problems, mental health, vitality, bodily pain and general health. The scores of the SF-36 scales can range between 0 and 100, with a higher score indicating a better functional health status. The Euroqol 5D is composed of 5 questions on different dimensions of quality of life: mobility, self-care, usual activities, pain/discomfort, anxiety/depression. Response options vary from 1 to 3, higher scores indicating worse quality of life. On an additional VAS score (0-100), patients rate their current health status, higher scores indicating better health status.
- Volume of OT, PT and ST utilization. Frequency of allied healthcare visits x total treatment duration was used to calculate volume. The procedure for obtaining these data is described in more detail below.
- Relative 'overuse' or 'underuse' of OT, PT and ST. The difference between actual allied healthcare utilization and the utilization as recommended by the multidisciplinary team, where the latter is considered the gold standard.
- Costs of PT, OT and ST. These were calculated using standard cost prices for allied health services (€ 23.60 per 30-min session) and consultant care (€98 per hour) in 2003, in accordance with national guidelines for cost calculation of health services[19].
- Patients' rating of the assessments and advice was evaluated with questionnaires (described in more detail below).

Volume of OT, PT and ST

In both groups the volume (number and duration of treatment sessions) of OT, PT and ST received was registered using patient questionnaires. At baseline, patients were asked to register the amount of OT, PT and ST received during the previous 6 months. In addition, patients in the reference group were asked to register the amount of OT, PT or OT during the subsequent 8 weeks prospectively. This amount was extrapolated to 6 months (26 weeks) to obtain comparable time periods. At six months follow up, the patients in the intervention group retrospectively registered the

amount of therapy received in the preceding 6 months. The number of allied healthcare visits was categorised as follows: no sessions, 1 to 10 sessions, 11 to 25 sessions, 26 to 52 sessions, and more than 52 sessions. The choice for the two latter categories was related to a treatment frequency of once a week (26 sessions in half a year) or twice a week (52 sessions in half a year).
To calculate the number of treatment sessions as recommended in the multidisciplinary reports, the following assumptions were made:

- The OT often advised one to five or five to ten sessions of OT, as it was difficult to know exactly how many sessions were needed. These were rated as five or ten sessions of 30 min, respectively;
- The PT regularly suggested to gradually decrease the PT to a lower frequency e.g. from twice a week to once a week or in some cases to once a month. In such cases, a gradual decrease of one session per month was assumed;
- If the PT advised a certain number of PT sessions followed by a re-evaluation, only the number of recommended sessions was used in the calculation;
- If hydrotherapy was suggested, this was counted as PT only if the need for PT coaching was specified.

Patients' rating of the assessments and advice
Patients in the intervention group were asked for their opinion on the assessments and advice for OT, PT and ST separately. They were asked whether the assessment had been valuable for them and why (not). The reasons for a valuable assessment included 'attention was paid to my problems and needs', 'new information or advice was given' or both. If the assessment was not considered valuable, the patient could specify why. These specifications included 'I do not experience problems or needs', 'I did not receive new information or advice' or other reasons. Patients were also asked whether or not they agreed with the advice given by the OT, PT and ST respectively.

Statistical analysis
An independent *t*-test was used to test whether there were significant differences in age between the two groups. A Mann-Whitney Test was used to test whether there were significant differences between the groups in medical diagnosis, duration of the complaints, generic health status and quality of life. Descriptive statistics (percentages) were used to describe the

amount of OT, PT or ST received, and differences between groups were tested using a Mann-Whitney test. A Wilcoxon test was used to test for significant differences between the amount of therapy received at baseline and at 6 months for both groups.

Results

Patients

For the reference group, medical records of 250 eligible patients were reviewed; 144 patients did not meet the inclusion criteria (Figure 2). The main reason was the absence of a probable or definite neuromuscular disease. Many patients were still in the diagnostic phase. Other reasons included age (younger than 18) or communication problems (insufficient command of the Dutch Language). Of the 106 patients included in the reference group, 87 returned the health care diary. For the intervention group, medical records of 257 eligible patients were reviewed; 155 patients were excluded for the same reasons as in the reference group. Of the 102 patients who participated, 88 returned the questionnaire reporting the amount of therapy received in the previous 6 months.

Baseline characteristics revealed no significant differences between the two groups in age, medical diagnosis, duration of the complaints, generic health status, quality of life or amount of OT, PT and ST received during the previous 6 months ($p>0.05$) (Tables I and II). The mean age was 47 years (SD 15) and 49 years (sd 13) for the reference group and intervention group respectively. The male/female distribution was 57/49 in the reference group 51/51 in the intervention group. In both groups all NMD categories (disorders of the motor neuron, motor nerve root, peripheral nerve, neuromuscular transmission and muscles) were included, with the majority suffering from a muscle disease (54% in the reference group and 67% in the intervention group). The duration of the complaints lasted a year or more in the majority of the patients in both groups (>95%). The SF-36 dimensions Physical functioning, Role limitations due to physical problems and General health had the lowest scores (mean scores below 50 with high standard deviations), indicating substantial, though variable physical problems in both groups.

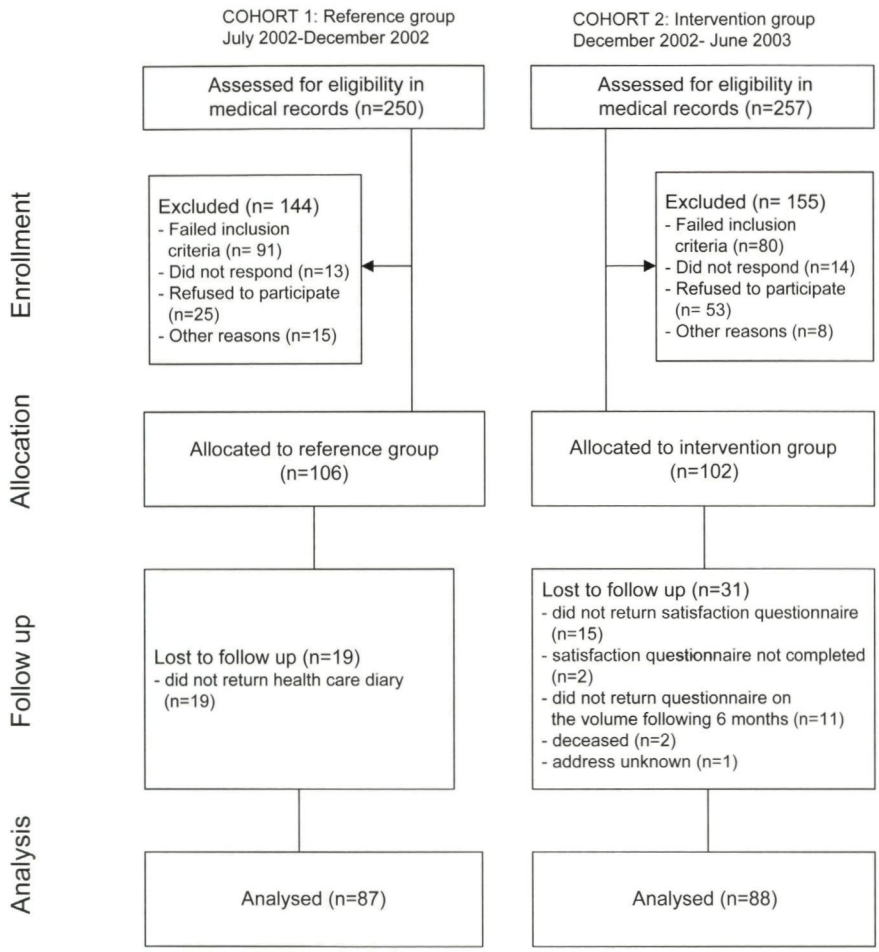

Figure 2. Inclusion and follow-up of patients.

Table I. Characteristics of the reference group and the intervention group

	Reference group, n=106	**Intervention group, n=102**
Age (years)		
Mean / standard deviation / range	47 / 15 / 20-83	49 / 13 / 20-73
Gender		
Male / female	57 / 49	51 / 51
Medical diagnosis		
Motor neuron disorders	9	8
Motor nerve root disorders	11	4
Peripheral nerve disorders	20	15
Neuromuscular transmission disorders	9	7
Muscle disorders	57	68
muscular dystrophy / myopathy / myositis	32 / 19 / 6	39 / 21/ 8
Duration of complaints		
0-6 months / 6-12 months / >12 months	2 / 5 / 99	3 / 4 / 95
Generic health status (SF 36) mean (SD)		
Physical Functioning	37 (26)	33 (27)
Social Functioning	70 (24)	68 (25)
Role limitations – physical	48 (41)	39 (39)
Role limitations – emotional	78 (39)	69 (42)
Mental health	74 (17)	72 (18)
Vitality	55(18)	51 (20)
Bodily pain	70 (28)	65 (30)
General Health	45 (21)	42 (22)
Quality of Life (EQ-5D) mean (SD)		
Mobility	1,9 (0,4)	1,9 (0,4)
Self-care	1,5 (0,6)	1,5 (0,6)
Usual activities	1,8 (0,5)	1,9 (0,4)
Pain/discomfort	1,8 (0,5)	1,9 (0,5)
Anxiety/depression	1,2 (0,4)	1,2 (0,5)
Health status	62 (17)	60 (18)

*$p<0.05$.

The problems in mental health or Role limitations due to emotional problems had the highest scores, indicating least problems in these areas. The Euroqol 5D showed similar results with problems in all dimensions, but the least in the dimension Anxiety/depression (Table I). The mean health status in the Euroqol was 62 (SD 17) and 60 (SD 18) for the reference group and intervention group, respectively.

The volume of OT, PT and ST at baseline was similar in both groups: Less than 10% had received a short period of OT; 32% (reference group) and 40% (intervention group) of the patients had received PT, most of them more than once a week; and there were hardly any patients that had had ST in the past 6 months (Table II).

Table II. Amount of therapy at baseline for the reference group and intervention group

	Reference group n=106	**Intervention group n=102**
Baseline Occupational Therapy		
0 sessions	96	93
1 – 10 sessions	7	8
11 – 25 sessions	2	1
26 – 52 sessions	1	0
> 52 sessions	0	0
Baseline Physical Therapy		
0 sessions	72	61
1 – 10 sessions	1	5
11 – 25 sessions	13	11
26 – 52 sessions	17	21
> 52 sessions	3	4
Baseline Speech Therapy		
0 sessions	101	101
1 – 10 sessions	5	0
11 – 25 sessions	0	1
26 – 52 sessions	0	0
> 52 sessions	0	0

*$p<0.05$

Multidisciplinary assessment and advice

After the OT assessments (n=102), 44 patients (43%) received advice for further treatment and 28 patients (27%) received a single advice. The PT assessments (n=102) resulted in an indication for further treatment in 46 patients (45%) and an indication for a single advice in 19 patients (19%). After the ST assessments (n=102), six patients (6%) were advised to have further ST and 41 patients (40%) received a single advice.

Patients' rating of the assessments and advice

Most patients valued the OT, PT or ST assessments highly (Table III), giving as main reason that they appreciated the attention paid to their specific problems and needs, or that they were given new information or advice.

Patients often mentioned both reasons. Other reasons for a positive assessment were that patients were pleased for having been examined so thoroughly and that they were relieved for still doing reasonably well.

Reasons for not appreciating the assessment were that the advice was difficult to implement or patients did not feel that their problems were taken seriously. At least 95% of the patients agreed with the advice given for OT (99%), PT (95%) and ST (100%).

Table III. Patient appreciation of their assessment

	Occupational Therapy (n=89)	**Physical Therapy (n=86)**	**Speech Therapy (n=88)**
Valuable			
• Because attention was paid to my problems and needs	40.4%	44.2%	28.4%
• Because new information or advice was given	10.1%	11.6%	12.5%
• Because of both of the foregoing reasons	32.6%	29.1%	28.4%
• For other reasons	0%	4.7%	3.4%
Total	83.1%	89.5%	72.7%
Not valuable			
• No problems nor needs	12.4%	2.3%	25%
• No new information nor advice	1.1%	3.5%	2.3%
• For other reasons	3.4%	4.7%	0%
Total	16.9%	10.5%	27.3%

Volume of OT, PT and ST

In the reference group, the volume of OT, PT or ST received during the 6 months before inclusion in the study (baseline) and at 6 months follow up were not significantly different (Table IV). In the intervention group, there was a significant underuse of OT in 40% of the patients and hardly any overuse of OT. The mean difference between the usual and recommended amount of OT was 2 sessions per patient in 6 months (underuse). There was a significant underuse of PT in 22% of patients and overuse in 32%, with a net mean difference of 6 sessions per patient (overuse). There was limited underuse and overuse of ST (Table V).

Table IV. OT, PT and ST received by the reference group in the 6 months preceding the study and in the subsequent 6 months

Number of treatment sessions	Reference group Previous 6 months (n=106)	Reference group Following 6 months (n=87)
*Occupational Therapy**		
0	90.6%	93.1%
1 - 10	6.6%	5.7%
11 - 25	1.9%	1.1%
26 - 52	0.9%	0%
> 52	0%	0%
Physical Therapy†		
0	67.9%	59.8%
1 - 10	0.9%	2.3%
11 - 25	12.3%	12.6%
26 - 52	16.0%	25.3%
> 52	2.8%	0%
Speech Therapy‡		
0	95.3%	97.7%
1 - 10	4.7 %	1.1%
11 - 25	0%	0%
26 - 52	0%	1.1%
> 52	0%	0%

*p>0.1 for the volume of OT; †p>0.05 for PT ; ‡p>0.7 for ST.

Table VI shows the number and distribution of therapy sessions received over 6 months in the intervention group. It was recommended that more patients should have a short course of OT, PT and ST. Compared with the amount of PT received at baseline, the multidisciplinary advice was amounted to a reduction of the number of PT sessions. Comparison of therapy received at baseline with that at 6 months follow up revealed a non-significant reduction of PT and a significant increase of OT and ST (Table VI). At baseline, fewer than 10% of patient had had a short course of OT; at 6 months, this number exceeded 25%. The number of patients receiving 1 to 10 sessions of PT increased from nearly 5% at baseline to more than 11% at 6 months. For ST, only 1% of patients had received a short course of ST at baseline, whereas this number rose to more than 10% at 6 months. The data for the 6-month follow-up showed that the therapy advice was only poorly implemented (Table VI).

Table V. Percentage of overuse and underuse of OT, PT and ST and the overall mean difference between amount of therapy received at baseline and the amount advised by OT, PT and ST (n=102)

	Occupational Therapy	**Physical Therapy**	**Speech Therapy**
Overuse	2%	32%	1%
Neutral	58%	46%	94%
Underuse	40%	22%	5%
Mean difference (Standard deviation)	2.1 (4.0) Under-use	6.1 (15.2) Over-use	0.4 (2.6) Under-use

Impact of the multidisciplinary advice on health care costs

The costs of the multidisciplinary assessments and integrated advice were assumed to consist of personnel costs only, and were estimated at €245 per patient. Each PT and OT assessment lasted an hour and the mean duration of the ST assessment was half an hour. Reports took about half an hour to write. Using the cost price of €23.60 for half an hour, the cost of the allied health assessment and reporting were 8 x 23.60 = €188.80. The multidisciplinary meeting lasted 10 minutes per patient, leading to a cost of €23.60 for the allied health professionals (OT, PT and ST) and €32.67 for the consultants (neurologist and rehabilitation physician).

Comparing the costs of current practice (OT, PT and ST at baseline) and the costs of the recommended therapy showed that recommended practice could lead to mean cost savings of €85.20 per patient (mean increase of 2.1 OT sessions and 0.4 ST sessions and mean decrease of 6.1 PT sessions per patient). However, at 6 months the recommendations were only partially implemented. While the mean amount of OT received had increased by 1.60 sessions per patient, the mean amount of ST received had increased by 0.1 sessions per patient and that of PT had decreased by 0.8 sessions per patient. The actual amount of OT, PT and ST received at 6 months increased costs by €21.24 per patient in addition of the costs of the multidisciplinary assessment and advice.

Table VI. Volume of OT, PT and ST in the intervention group during the 6 months preceding study entry, based on the advice and at 6 months

Number of treatment sessions	Intervention group at baseline (n=102)	Intervention group based on advice (n=102)	Intervention group at 6 months (n=88)
OT sessions	63	281*	177†
0	91.2%	55.9%	72.7%
1 - 10	7.8%	44.1%	22.7%
11 - 25	1.0%	0%	2.3%
26 - 52	0%	0%	2.3%
> 52	0%	0%	0%
PT sessions	1333	708*	1011
0	59.8%	51%	58.0%
1 - 10	4.9%	24.5%	11.4%
11 - 25	10.8%	13.7%	9.1%
26 - 52	20.6%	9.8%	19.3%
> 52	3.9%	1.0%	2.3%
ST sessions	12	48	23†
0	99%	94.1%	88.6%
1 - 10	0%	3.9%	10.2%
11 - 25	1.0%	2.0%	1.1%
26 - 52	0%	0%	0%
> 52	0%	0%	0%

* $p<0.01$ for the volume of OT ($p=0.00$), PT ($p=0.00$) and ST ($p=0.17$) retrospectively measured compared to the volume based on the advice. † $p< 0.05$ for the volume of OT (0.01), PT (0.55) and ST (0.05) retrospectively measured at baseline compared to the volume retrospectively measured at 6 months follow up.

Discussion

In this study we compared the volume of OT, PT and ST in NMD currently received with the volume recommended by an expert multidisciplinary team. In the absence of scientific and solid empirical evidence (a gold standard) regarding the most appropriate volume of allied health care for patients with NMD, this 'expert advice' was considered as 'next-best' standard of reference.

Our results show that the expert advice regarding the volume of OT, PT and ST differed considerably from the actual service provision among patients with NMD. According to the expert advice, far more patients would need a short course of therapy or advice during a single consultation. In this article we use the terms 'overuse' of 'underuse' to describe the difference between the actual volume of OT, PT and ST and the recommended volume.

For sake of brevity, we have used these terms in our paper, although these terms imply a standard of care that is presently unavailable.

Regarding OT, there was a substantial 'underuse' of OT in current practice compared to the recommended volume of OT. This underuse is confirmed in an inventory of allied health care for chronically disabled in the Netherlands[20]. An unmet need for OT among patients with diseases of the nervous system is reported. Reasons for this unmet need include: 1) lack of awareness in patients and referring physicians of what OT has to offer; 2) problems with referral (too late or not at all) in one third of the patients; 3) problematic OT funding by health insurance companies for about 46% of chronic conditions; and 4) the poor availability of OT in primary care[20]. These reasons may also explain the difficulties experienced to implement the expert advice.

Compared to the recommended volume of PT, there was 'overuse' as well as 'underuse' of PT in current practice. PT overuse was often the result of a higher frequency and duration of PT in current practice than suggested in the expert advice. Instead of continuous PT, a short course of PT was advised to address specific treatment goals, such as instructions to the patient how to maintain an optimal physical condition with an individual exercise programme. It appears that patients are insufficiently aware that they can continue exercises on an individual basis, not necessarily requiring professional supervision[20]. In our study often 6-8 PT sessions were advised, after which patients were encouraged to further carry out the exercises by themselves. A comparable PT approach, with 6-8 sessions, was described in a study on the implementation of clinical guidelines for patients with low back pain[21].

In current practice there was also 'underuse' of PT. This was the case when patients did not have PT, but the multidisciplinary team recommended that they received instruction and advice within a single consultation or within a short episode of PT. The Dutch inventory of allied health care reports that it is difficult to generalise about the need for PT in chronic conditions and that the duration of therapy depends on the results of therapy, the age of the patient and the type of health problems[20]. Patient organizations have pointed out that physiotherapists have insufficient knowledge concerning more than half of the 74 chronic conditions for which PT is indicated. Also physiotherapists themselves feel that they lack expertise in 70% of chronic conditions. Besides, in 30% of the chronic conditions for which PT is indicated, the coordination of the care of different professionals

is considered a problem[20]. These findings support the need of an integrated multidisciplinary approach.

There was some 'underuse' of ST when comparing the volume of ST recommended in the expert advice with current volume of ST. A considerable number of patients received information, instruction and advice on problems with eating and drinking and communication during a single ST consultation. Generally an unmet need for ST is estimated in patients with chronic progressive neurological conditions with voice or speech disorders, hearing disorders, breathing problems and generally communication problems[20]. It is remarkable that problems with swallowing or chewing (eating and drinking) are not reported in the Dutch inventory of allied health care. Patients with chronic progressive neurological conditions such as NMD regularly experience such problems. The speech therapists working at the neuromuscular centre of Nijmegen have specific expertise in the diagnosis and provision of advice regarding these problems[22-24].

Patient organizations and speech therapists both feel that generally speech therapists lack knowledge about chronic conditions. Besides, patients are not aware of what ST has to offer and referring physicians lack insight into when a patient should be referred for ST[20].

Our findings and the reported problems regarding OT, PT and ST for chronic conditions[20], support the value of a integrated multidisciplinary expert advice regarding appropriate allied health care for patients with NMD. Also, the majority of the patients rated the multidisciplinary assessments and advice positively.

Obviously, the implementation of treatment recommendations could be improved. Reasons for poor implementation include limited awareness among patients and referring physicians, and problems with referral, availability and funding. Additionally, there may be problems with the feasibility of the advice, limited motivation to change routines or limited knowledge and skills of therapists to implement the advice. These causes have been explored and will be analysed and published together with possible ways to obtain a stronger commitment on the part of allied health services in primary care.

Although the multidisciplinary assessment resulted in a decrease in the cost of allied health services, these savings were offset by the extra costs incurred by the multidisciplinary assessments. Clearly, ways need to be found to obtain a more favourable balance between the costs and savings of the multidisciplinary approach.

Limitations of this study include possible recall bias of the volume of allied health care. Still, we preferred patient questionnaires above patient records since the latter did not provide reliable information on this subject.

Secondly, one could question whether service utilisation during an eight week period may be extrapolated to 26 weeks. However, the amount of OT, PT and ST measured retrospectively with a patient questionnaire did not differ significantly with the amount measured prospectively with a health care diary for eight weeks and extrapolated to six months. We therefore felt that it was justifiable to conclude that the amount of OT, PT and ST did not change in the usual care group.

On the basis of our findings, we cannot comment on the effect of the intervention on patients' health status in terms of functional status or quality of life, because this would require a different study design and different outcome measures. Such studies are very much needed.

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Part I: Professional perspective

Chapter 3
Implementation of multidisciplinary advice to allied health care professionals regarding the management of their patients with neuromuscular diseases

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Abstract

Purpose: Patients with neuromuscular diseases (NMDs) do not always receive appropriate allied health care. This is partially because of the large heterogeneity among these conditions, some of which are quite rare. Individual allied health care professionals, therefore, have relatively little opportunity to develop relevant experience with these patients. To overcome this problem, we developed specialist multidisciplinary advice regarding management of patients with NMD for occupational therapy (OT), physical therapy (PT) and speech therapy (ST) in a primary care or rehabilitation setting. The aims of the study were to explore to what extent this allied health care advice was implemented with a focus on the amount of therapy received and to explore possible barriers to implementation.

Methods: One-hundred two patients visited the Neuromuscular Centre Nijmegen for OT, PT and ST consultations. Integrated allied health care advice was written on the basis of these consultations and a multidisciplinary meeting. All patients, their therapists and rehabilitation physicians received this advice. Following the advice, questionnaires were sent out at baseline and at 6 months follow up, collecting data on implementation of the amount of therapy suggested and on possible barriers for adherence.

Results: Advice for ST and OT was fully implemented in primary care, but only partially (58%) in a rehabilitation setting. Advice to reduce the amount of PT was implemented in only 15% of the cases. Possible barriers were related to the advice itself (feasibility of treatment duration, correctness and completeness), the patient (motivation) and the professional (experience in treatment of NMDs). Therapists expressed a desire to have the opportunity to discuss the treatment advice with the multidisciplinary team.

Conclusions: The extent to which multidisciplinary advice was implemented differed for OT and ST compared to PT and for the setting (primary care or rehabilitation). Possible barriers were identified at different levels. We recommend follow-up telephone calls to provide therapists opportunity for discussion.

Introduction

At least one in 3500 individuals develops a disabling neuromuscular disease (NMD) at some time during childhood or later in life[1]. Sooner or later, these patients are confronted with impairments in functions, activity limitations and participation restrictions. When this is the case, they may benefit from allied health care, including occupational therapy (OT), physical therapy (PT) and speech therapy (ST). Unfortunately, such therapy is not always adequately provided to meet the complex needs of this patient population[2-5]. This may result from the fact that patients and referring physicians are not fully aware of existing treatment opportunities, the weak evidence for OT, PT and ST in NMD[6-8], and the scarcity of therapists in primary care who have sufficient clinical experience with these patients[4;5]. These findings support the need for specialist services for patients with NMD. Both in the UK[5] and the Netherlands, care for patients with NMD is concentrated in a limited number of specialised centres. However, because of limited capacity, it is impracticable to manage all patients under treatment in these centres. For patients to fully benefit from such services, knowledge and experience should be transferred to OTs, PTs and STs in primary care and rehabilitation centres, where their treatment can be continued.

For this purpose, our neuromuscular centre has developed a consultation and advice service for OT, PT, and ST. Part of this service consists of the development of integrated, multidisciplinary allied health care treatment advice for each individual patient. This advice is based on the best available scientific evidence and clinical experience. For OT, scientific evidence consists of indications for the efficacy of hand training in muscular diseases[7]; for PT, there is weak evidence of efficacy for strengthening exercise in combination with aerobic training in different types of NMD and for breathing exercises in some specific forms of NMD[6]; for ST, evidence indicates that correction of head position in patients with OPMD improves swallowing[8]. There is no evidence of efficacy of interventions such as education of patients and family, practicing skills or activities, or the use of assistive advices. Also, lifestyle modification strategies including pace setting or other energy conservation techniques have not yet been studied for their effectiveness. For best practice though, these strategies are used in OT, PT and ST in order to enable participation in activities and roles which are meaningful to the daily life of persons.

The OT, PT and ST consultations included assessments of body functions, activities and participation[2]. Upon completion of the assessment,

some patients received advice with no further treatment required. If further treatment was advised, the objectives, the content and the expected duration were explicitly stated[2]. An example of treatment objectives for PT was 'to increase stamina to be able to cycle for at least half an hour and to implement regular exercise in daily life'. To reach these objectives, aerobic exercise on a bicycle was suggested with a frequency of twice a week for 6 weeks followed by an evaluation. For OT, an example of an objective was 'to have enough energy to make dinner safely without fear of falling'. To reach this goal, supporting the patient in the use of energy conservation strategies and assistive devices was suggested. The expected duration was 5 sessions of OT in the home situation. Regarding ST, many patients received advice during the consultation and did not need further treatment. These activities involved for instance speaking slower, eliminating certain food or eating and drinking with slightly flexed head position.

The advice was offered to the OTs, PTs or STs in primary care or to a rehabilitation physician. However, following such advice may require changes in established long standing practices in the management of these patients, hampering implementation[9]. Therefore, the aims of this study were:

1. to investigate the extent to which the allied health care advice was implemented in OT, PT and ST practice in primary care and rehabilitation setting and
2. to identify possible barriers for implementation, based on a conceptual model of implementation of changes in health care[9]; these barriers were related to the advice itself, the patient, the professional, and the organisation.

Methods

Patients

A prospective cohort study was carried out with consecutive patients visiting the outpatient department of the Neuromuscular Centre Nijmegen, the Netherlands. Patients were asked to participate if they fulfilled the following inclusion criteria: age 18 years or older; a definite or probable NMD; sufficient command of the Dutch language and informed consent to participate in this study. The study was approved by the local medical ethics committee.

Procedure and questionnaires

The procedures that were used in this study are schematically represented in Figure 1. Eligibility for inclusion was assessed on the basis of a medical record review. After receiving written informed consent, patients were invited to attend the Neuromuscular Centre Nijmegen twice. At the first visit (T0), they consulted a neurologist and completed *Patient questionnaire 1*, which asked the patient about the use of allied health care during the previous 6 months, their medical diagnosis and duration of the disease. Patients reporting to have OT, PT or ST, were asked to hand their therapist *Professional questionnaire 1,* asking information regarding frequency, duration and content of current therapy. These therapists were also asked to estimate the number of patients with NMD they treat annually, and whether they would appreciate advice from a centre of expertise. Within 2 weeks from the first visit, patients were seen by an OT, PT and ST from the centre. Integrated allied health care advice was developed on the basis of these consultations and a multidisciplinary meeting (2). The advice was sent to the patients together with *Patient questionnaire 2* (T1), asking for their opinion on the OT, PT and ST consultations and their satisfaction with the advice provided (2). The therapists in primary care also received this advice as well as *Professional questionnaire 2*. This questionnaire included statements about the advice (clarity, feasibility and credibility), about the professionals' appreciation of the advice and the organisation and the communication around the advice (see Appendix 1). Response options for each statement were 'agree', 'neither agree nor disagree', or 'disagree'. In case patients had been treated by a rehabilitation physician prior to the consultation, or if the advice involved a multidisciplinary approach to be coordinated by a rehabilitation physician, the advice was sent to the rehabilitation physician.

After six months (T2), *Patient questionnaire 3* and *Professional questionnaire 3* were sent to the patients and their allied health care providers or rehabilitation physicians. Patients were questioned about the amount of OT, PT or ST services that they had received since the advice. Allied health care professionals or rehabilitation physicians were questioned about the advice itself (clarity, feasibility, credibility, adaptability), about the patient (motivation), about themselves as a professional (appreciation, knowledge/skills, attitudes/ habits, communication) and about the process (referral, practice, time, waiting list, funding and communication) (see Appendix 2).

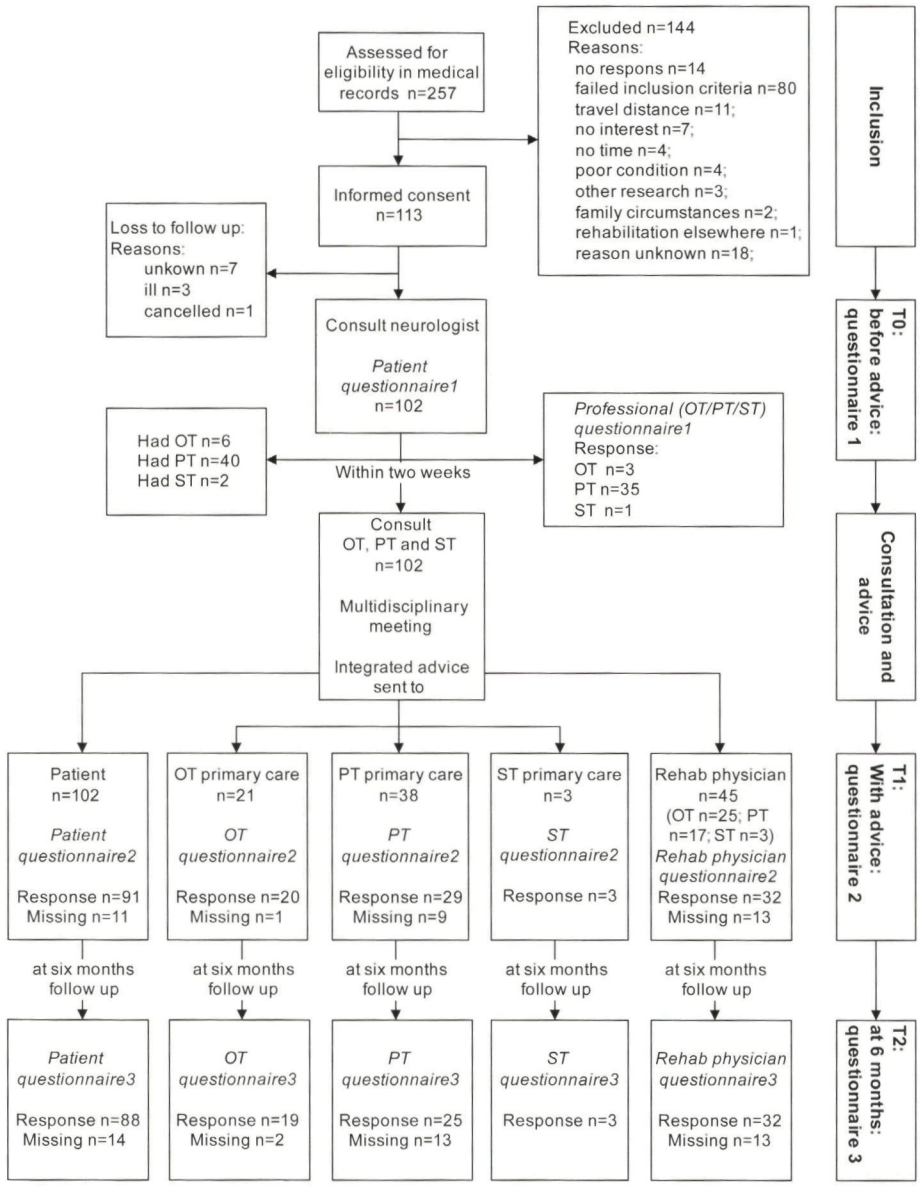

Figure 1. Flowchart showing the inclusion, consultation and advice, and follow up of patients and professionals involved.

Implementation measure

For OT and ST, implementation was considered successful if patients had indeed received OT or ST as recommended. Patients who had received advice for OT or ST were divided into two groups: patients who were referred directly to OT or ST in primary care and patients who were referred to a rehabilitation physician. For PT, this distinction did not apply, since most patients already had PT in primary care and also visited a rehabilitation physician. For PT, implementation was considered successful if patients received the recommended amount of PT as advised. The outcomes were divided into five categories: continued *not* to have PT, discontinued PT, reduced the amount of PT, continued the same amount of PT, or initiated PT as recommended.

Statistical analysis

Chi-square statistics were carried out as appropriate to test the significance of the correlation between referral procedure (primary care or rehabilitation setting) and successful implementation for OT and ST and the correlation between the category regarding the volume advised and implementation for PT.

For the analysis of *Professional questionnaires 2* and *3* descriptive statistics were used. The percentages 'agreement', 'neither agreement nor disagreement' and 'disagreement' were calculated for each statement. If less than 70% of the respondents agreed (disagreed when negatively formulated) with a statement, these statements were further analyzed with use of non-parametric (Chi-square) statistical test procedures.

Results

Inclusion and follow up of patients

The process of inclusion, baseline measurements, consultation, advice and follow up measurements are presented in Figure 1. In total, 257 medical records were reviewed to identify eligible patients. Eighty patients failed the inclusion criteria and another 64 patients did not participate for a variety of reasons. Informed consent was obtained from 113 patients. Eleven patients dropped out, leaving 102 patients who participated in the research project and who completed *Patient questionnaire 1*. The characteristics of the patients including age, gender, medical diagnosis and duration of disease are presented in Table I.

Table I. Characteristics of patients included (n=102)

Age (years)	
Mean (standard deviation)	49 (13)
range	20-73
Gender	
Male / female ratio	51 / 51
Medical diagnosis	
Motor neuron disorders	8
Motor nerve root disorders	4
Peripheral nerve disorders	15
Neuromuscular transmission disorders	7
Muscle disorders	68
muscular dystrophies	39
myopathies	21
myositis	8
Duration of complaints	
0-6 months	3
6-12 months	4
>12 months	95

All patients (n=102) visited the OT, PT and ST of the neuromuscular centre and received written integrated advice together with *Patient questionnaire 2*. The response rate for *Patient questionnaire 2* was 89% (n=91). Most patients (83% for OT, 90% for PT and 73% for ST) highly valued the assessments and appreciated the professional attention paid to their problems and needs. Where patients indicated they did not appreciate the assessment and advice, the main reasons for this were that they did not experience any problems or did not have any needs in this particular area. Nearly all patients agreed with the advice given for OT (99%), PT (95%) and ST (100%)[2]. The response to *Patient questionnaire 3* was 86% (n=88) (Figure 1).

Implementation of advice on occupational therapy

During the 6 months prior to the consultations, nine patients had had OT, of which six patients still received OT. Forty-five percent of the patients (n=46) received advice for OT treatment, 21 (46%) of whom were referred directly to primary care and 25 (54%) to a rehabilitation physician (Figure 1). The percentage of successful implementation for patients referred to OT in primary care was 100%, versus 58% for patients referred to a rehabilitation physician ($p<0.001$). Reasons for not implementing the advice are presented in Table II. The reasons from the patients' perspective generally correspond

with the reasons from the perspective of the rehabilitation physician and include barriers at the level of the advice (unclear), the patient (personal reasons), the professional (disagreement with advice) and the organization (waiting list).

Table II. Reasons for not having had OT according to patients and rehabilitation physicians (n=10)

Patient's perspective	Rehabilitation physician's perspective
Did not know what to do	Patient never made an appointment
Illness of partner	Patient is not motivated
Appointment in near future	There is a waiting list for appointments
Rehabilitation physician disagreed	Problems had been addressed before (n=3)
No need for OT anymore	Questionnaire not completed (n=2)
OT advice integrated in PT	Patient is not in treatment anymore
Not specified (n=4)	Rehabilitation physician does not work here anymore
	Missing (n=2)

OT=occupational therapy; PT= physical therapy

The response rate for *OT questionnaire 2* was 95% (n=20) (see Figure 1). Less than 70% of OTs in primary care agreed that they would adopt the advice regarding the location where the treatment should be given. Specifically, OTs often preferred to treat a patient at home, when the advice recommended treatment in an OT department.

The response rate for *OT questionnaire 3* at 6 months follow up was 90% (n=19) (Figure 1). Less than 70% of OTs in primary care agreed with feasibility of all therapy goals (59%) or with the statement that therapists from the neuromuscular team were easily to contact (47%). In addition, 33% of OTs indicated lack of waiting lists and 67% indicated problems related to appropriate funding.

Implementation of advice on physical therapy

Forty patients (39%) had PT when they participated in the study. Thirty-five PTs (88%) returned *PT questionnaire 1* (Figure 1). The majority of the PTs (71%) treated patients for a duration of at least 1 year and 82% saw their patients at least once a week or more. Most PTs in primary care (73%) treated less than five patients with a NMD per year and all PTs appreciated receiving advice from the neuromuscular centre.

According to the multidisciplinary team, 45% of the patients had an indication for PT. Of these, 70% (n=32) were, in fact, under PT treatment.

Among patients who were considered not to have an indication for PT (n=54), 15% (n=8) were under PT treatment. For 63% of the patients (n=64) the advice did not involve any change (continue not to have PT or continue to have PT as given), and the advice was almost fully implemented. In contrast, implementation of advice recommending a change in the amount of PT was significantly less ($p<0.001$). Implementation of advice recommending a reduction in the amount of PT was 15% (2 out of 13), recommendations to discontinue PT were implemented in 37.5% (3 out of 8) and recommendations to initiate PT were implemented for 29% of the patients (2 out of 7) (Figure 2).

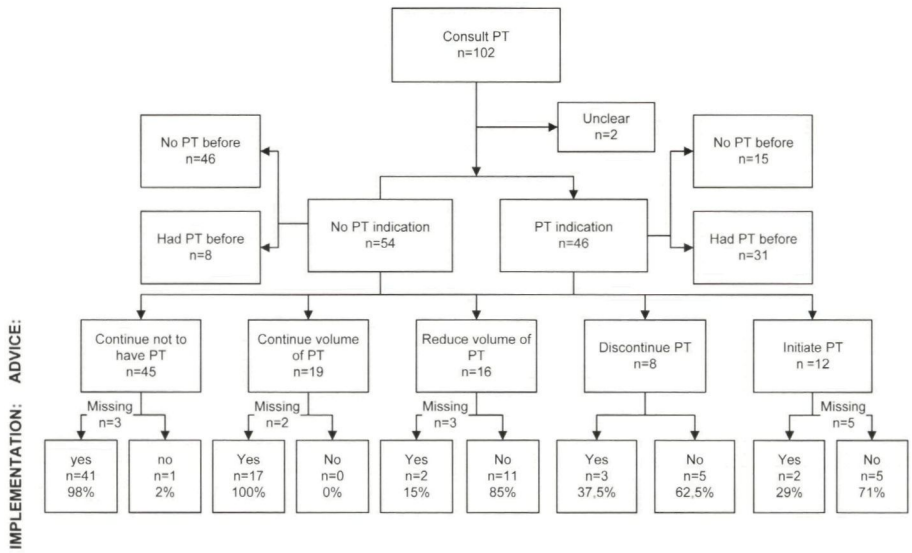

Figure 2. Flow chart implementation physical therapy advice. PT, physical therapy.
* For one patient there was an indication for PT only if the complaints did not disappear within two weeks and for another patient the advice was to discontinue PT in primary care and wait for the multidisciplinary rehabilitation programme.

The response rate for *PT questionnaire 2* was 76% (n=29) (Figure 1). Less than 70% of the PTs agreed with the six statements: clarity of treatment location, feasibility of therapy duration, feasibility of treatment location, completeness of advice, ability to provide information and opportunity for discussion. None of these statements correlated significantly with implementation of the advice.

The response rate for *PT questionnaire 3* was 66% (n=25) (Figure 1). Less than 70% of the PTs agreed with the 12 statements. These were statements regarding feasibility of the advice (treatment goals, treatment duration and treatment location), credibility of the advice (correctness of advice, completeness of information and need for more information), patient motivation, experience in treatment of patients with NMD, communication with the neuromuscular team (therapists easily to contact, referral received and reporting findings to referring physician), and appropriate funding. None of the statements had a significant impact on implementation (Chi-square test statistic $p>0.05$). Six statements, however, did seem to be related to implementation of the advice, including feasibility of the treatment duration, correctness of the advice, completeness of information, need for more information, experience with treatment of patients with NMD and patient motivation (Appendix 2).

Implementation of advice on speech therapy

Two out of 102 patients had ST in the 6 months prior to the multidisciplinary consultation (see Figure 1). One ST returned *ST questionnaire 1*. This ST saw less than five patients with NMD per year and valued advice from the neuromuscular centre. Six percent of the patients (n=6) received ST advice for further treatment. For three patients the advice was sent to a ST in primary care and for three patients the advice was sent to a rehabilitation physician (Figure 1). Four patients (67%) responded to *Patient questionnaire 2* and *3*. All STs responded to *ST questionnaire 2* and *3* and all rehabilitation physicians had returned the *Rehabilitation physician questionnaire 2* and *3*. All ST advice was fully implemented. Because of the limited number of questionnaires, no further analyses were undertaken.

Discussion

Our main finding was a wide difference between the implementation of advice for OT and ST and that of PT. OT and ST advice was fully implemented in primary care, which is remarkable, given that passive dissemination of information (only sending the advice) is generally an ineffective strategy[9]. Our high rate of implementation is very likely related to the current underuse of OT and ST (2), which, in turn, may result from the fact that OT and ST services are generally less well-known to patients and physicians, and the scarcity of scientific evidence for their effectiveness[6-8].

However, implementation was considerably less successful (58%) when the advice was sent to a rehabilitation physician. Barriers to implementation were identified at all levels (advice, patient, rehabilitation physician, and organisation). In most cases, the patients were known to the rehabilitation physician and the problems were generally more complex than the problems of the patients referred to primary care.

Implementation of advice to change the amount of PT was extremely poor. Potential barriers for implementation included feasibility of the suggested treatment duration, correctness of advice, completeness of information, need for more information and lack of opportunity for discussion about the treatment advice. Other factors that may have contributed to poor implementation include lack of patient motivation, PT's lack of clinical experience and knowledge with NMD and income deprivation[2;4].

It should be noted that NMD are chronic progressive conditions, with increasing loss of abilities. Although there is limited evidence regarding the effectiveness of PT on improving bodily functions, patients may perceive a need to control this gradual process of deterioration and may feel reassured having PT. This need for continuous monitoring of their condition by PT might have contributed to the limited implementation of advice to reduce the amount of PT. Other reasons might have been that the goals had not been reached within the timeframe suggested or that of new goals arose.

For an improved understanding and resolution of barriers related to feasibility, lack of information and correctness of information, a follow-up telephone contact with the professionals in primary care or rehabilitation setting would be a logical intervention strategy. A follow-up telephone call provides an opportunity to exchange experience, to provide additional information and available scientific evidence if needed to support the professionals in the decision making process. Another way to increase understanding of the barriers for implementation would have been a focus group with PTs to discuss the findings in respect to poor implementation of the amount of therapy advised.

A limitation of this study is that we could not assess whether the content of the advice was implemented. Nor did we evaluate the advice with patient-centred measures. Another shortcoming is that we investigated the implementation and possible barriers for all NMDs together. Because of small numbers for the various NMD diagnoses, we could not investigate whether implementation differed across the various diagnoses.

The aim of this study was to improve allied health services for patients with NMD by providing advice from a neuromuscular centre. This way, patients can continue to have allied health care close to where they live, and therapists are provided with knowledge and experience from a neuromuscular centre. To obtain greater uniformity in content of therapy, the accumulative knowledge and experience should precipitate into guidelines. We also strongly recommend the use of more explicitly defined goals of therapy and the use of instruments to measure change. This would provide patients and therapist with opportunities to discuss continuation or discontinuation of therapy.

Conclusions

We explored the extent to which multidisciplinary advice was implemented in daily care. Recommendations were generally readily adopted when additional therapies were suggested to address patient needs and where no capacity problems existed. Where we recommended critically evaluating, and possibly reducing therapy, this was hardly implemented. To improve implementation of this type of knowledge transfer, we suggest improved communication between professionals from the centre of expertise and local professionals, for instance through follow-up telephone calls. Also the use of agreed moments to evaluate therapy may assist in deciding when to continue and when to discontinue therapy.

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Appendix 1: Statements included in *Professional questionnaire 2*

Clarity of the advice in general

- The lay-out of the advice is clear
- The language of the advice is understandable
- The length of the advice is good

Clarity of specific OT/PT advice

- The OT/PT/ST advice is clear
- The language of the OT/PT/ST advice is understandable
- The length of the OT/PT/ST advice is good
- The therapy goals are clearly described in the OT/PT/ST advice
- The number of hours are clearly described in the OT/PT/ST advice
- The therapy frequency is clearly described in the OT/PT/ST advice
- The therapy duration is clearly described in the OT/PT/ST advice
- The place of therapy is clearly described in the OT/PT/ST advice

Feasibility of the advice

- The therapy goals are applicable
- The therapy goals are attainable
- The therapy frequency seems feasible
- The therapy duration seems feasible
- I will treat at the suggested place of therapy

Credibility of the advice

- The whole advice is complete
- The OT/PT/ST part is complete

Appreciation by the professional

- I appreciate the advice
- The advice is important
- The advice is a valuable contribution to the therapy

Organization and communication around the advice

- I have received an information leaflet
- The information leaflet was clear
- I was able to provide information
- There was opportunity for discussion.

OT= Occupational Therapy; PT= Physical Therapy; ST= Speech Therapy

Appendix 2: Statements included in *Professional questionnaire 3*

Clarity of the advice in general
- The advice is clearly formulated

Feasibility of the advice
- Certain therapy goals are not attainable
- The treatment duration is not feasible
- The treatment location is not feasible

Credibility of the advice
- Parts of the advice are incorrect
- The advice can be used in a wrong way
- The advice lacks important information
- I need additional information

Adaptability of the advice
- There is enough room for my own considerations
- There is enough room for the patients' wishes

Patient motivation
- The patient is not motivated for this advice

Appreciation by the professional
- I appreciate the advice
- I feel resistance towards the advice

Knowledge/skills of the professional
- I have enough knowledge to implement this advice
- I have enough skills to implement this advice
- I lack experience in the treatment of neuromuscular diseases

Attitudes/habits of the professional
- This advice resembles my working routines
- I have problems changing my routines

Communication of the professional
- I have discussed the advice with the patient
- I have reported my findings back to the referring physician

Organization and communication
- I have received a referral
- I have received the advice
- I have received an information leaflet
- Therapists were easily to contact
- The advice costs more time than I can give
- My practice is not suitable
- There is a waiting list of more than a month
- There is no appropriate financial funding.

Part II: Scientific perspective

Chapter 4
The evidence for occupational therapy for adults with neuromuscular diseases: a systematic review

Published as:
Cup EHC, Sturkenboom IHWM, Pieterse AJ, Hendricks HT, van Engelen BGM, Oostendorp RAB, van der Wilt GJ. (2008). The evidence for occupational therapy for adults with neuromuscular diseases: a systematic review. *OTJR: Occupation, Participation and Health*; 28(1):12-16.

Abstract

A systematic review was conducted to assess the evidence base of occupational therapy for adults with neuromuscular diseases. A comprehensive search for controlled and uncontrolled studies was performed in the Cochrane, Medline, CINAHL and Embase databases. After screening 3,534 titles and abstracts, six articles were retrieved for more detailed evaluation. Only two of the studies met all of the predefined criteria and were subjected to methodological assessment. These studies provide indications for the efficacy of hand training in muscle diseases. There is a lack of scientific evidence regarding the question of whether patients with neuromuscular diseases are better capable of participating in meaningful activities and roles through occupational therapy. Recommendations for future research are given.

Introduction

Neuromuscular diseases include disorders of the muscle, the neuromuscular transmission, the peripheral nerve, the nerve root, or the motor neuron. Most of these diseases have a slowly progressive course with increasing impairments such as muscle weakness, exercise intolerance, fatigue or pain, that ultimately result in limitations in activities and restrictions in participation. On the basis of these sequelae, it is conceivable that a considerable portion of these patients may benefit from occupational therapy. Indeed, in a cohort of 102 consecutive patients with neuromuscular diseases attending our outpatient clinic, 43% were considered to potentially benefit from occupational therapy[1]. However, this judgment was entirely based on the clinical expertise of the participating occupational therapists. The purpose of this article is to assess whether there is evidence for occupational therapy for patients with neuromuscular diseases.

Methods

We searched the Cochrane Database of Systematic Reviews and the Cochrane Central Register of Controlled Trials (The Cochrane Library 2005, Issue 3), Ovid Medline In-Process & Other Non-Indexed Citations and Ovid Medline (1966 through September 2005), WebSPIRS 5.1 CINAHL (1982 through September 2005) and WebSPIRS 5.03 EMBASE (Rehabilitation and Physical Medicine) (1995 through September 2005). The search strategy was based on: 1) a search of randomized controlled trials and controlled clinical trials as recommended by the Cochrane Neuromuscular Disease Group; 2) a search of other research designs; 3) a search of different types of neuromuscular diseases; and 4) a search of occupational therapy interventions. For all search strategies medical subject headings (MeSH) or indexed terms were used in addition to free text words. The full search strategy can be found in appendix A at the end of this chapter. Reference lists of selected articles and related articles were scanned for other potential relevant articles.

Selection criteria

Inclusion of articles was restricted to peer-reviewed articles with an abstract that were published in the English, German, French or Dutch language. Regarding types of studies, we included randomized controlled trials, controlled clinical trials and other designs such as pre-post designs. Single case studies were excluded.

Regarding the types of participants, we included studies with adults (individuals older than 18 years) with different types of neuromuscular diseases. Children or adolescents (individuals younger than 18 years) with Duchenne muscular dystrophy, spinal muscular atrophy or Becker muscular dystrophy were not included. The content and the context of occupational therapy in children are different from occupational therapy for adults with neuromuscular diseases due to aspects such as development and growth.

Entrapment neuropathies (such as carpal tunnel syndrome or cubital tunnel syndrome), radiculopathies, thoracic outlet syndrome or diabetic neuropathies were excluded, because we were interested in the chronic progressive types of neuromuscular diseases. Diagnoses with impairments similar to those in neuromuscular diseases, such as chronic fatigue syndrome, spinal cord injuries, reflex sympathetic dystrophy or complex regional pain syndrome, forms of cancer, and acquired immunodeficiency syndrome, were also excluded.

Occupational therapy interventions had to include training of activities of daily living (including self care, mobility, work, household tasks, or leisure) skills training (including fine motor skills), advice and instruction in the use of assistive devices, provision of splints and slings, counseling on energy conservation strategies, educating patient, families and caregivers, or a combination of the above. Studies evaluating specific assistive devices such as wheelchairs, robotic arms or mobile arm supports were excluded unless the evaluation comprised instruction and advice specifically provided by occupational therapists.

Primary outcome measures had to be at the level of activities and participation, quality of life, or general health. Outcome measures at the level of pain, fatigue, muscle strength or fine motor skills (dexterity) were accepted as secondary outcome measures.

Procedure for inclusion

A preliminary screening of titles and abstracts was performed by two qualified occupational therapists (EHCC and IHWMS). Full texts of studies, that were considered potentially eligible for inclusion, were reviewed further. Based on the full articles, the two reviewers independently made a final selection using the criteria described above. In cases of different conclusions, consensus was reached through discussion.

Methodological Quality

The methodological quality of the studies was independently rated using a selection of the list with criteria recommended by van Tulder et al.[2]. Because it is not possible for the therapist or the patient to be blinded for an occupational therapy intervention, these criteria were discarded. Therefore, the adapted list consisted of nine criteria for internal validity, six descriptive criteria and two statistical criteria for randomized controlled trials and controlled clinical trials (Table 1). All criteria were scored as yes, no, or not clear. Equal weight was given to all items.

Randomized controlled trials and controlled clinical trials were considered high quality if at least six of nine internal validity criteria, three of six descriptive criteria, and one of two statistical criteria were scored positively[3]. Other research designs were considered sufficient quality if four of nine internal validity criteria, two of six descriptive criteria, and one of two statistical criteria were scored positively[3]. The two reviewers independently assessed the methodological quality and disagreements were resolved by discussion.

Data analysis

A standardized data extraction form was used to present details of included studies, such as first author, year of publication, number of patients, patient characteristics (inclusion criteria, gender and age), description of outcome measures, intervention (intensity and content), and results. A best-evidence synthesis was presented by attributing specific levels of evidence to the various intervention strategies, taking into account the design of the studies, the methodological quality, the type of outcome measures and the statistical significance of the findings[3].

Results

The initial search strategy resulted in a total of 3,534 citations. After screening by title and abstract, the majority of the studies (n=3,528) did not meet the predefined criteria for the type of study, type of participants, type of intervention, or type of outcome measures. Six full-text articles were retrieved for detailed evaluation[4-9].

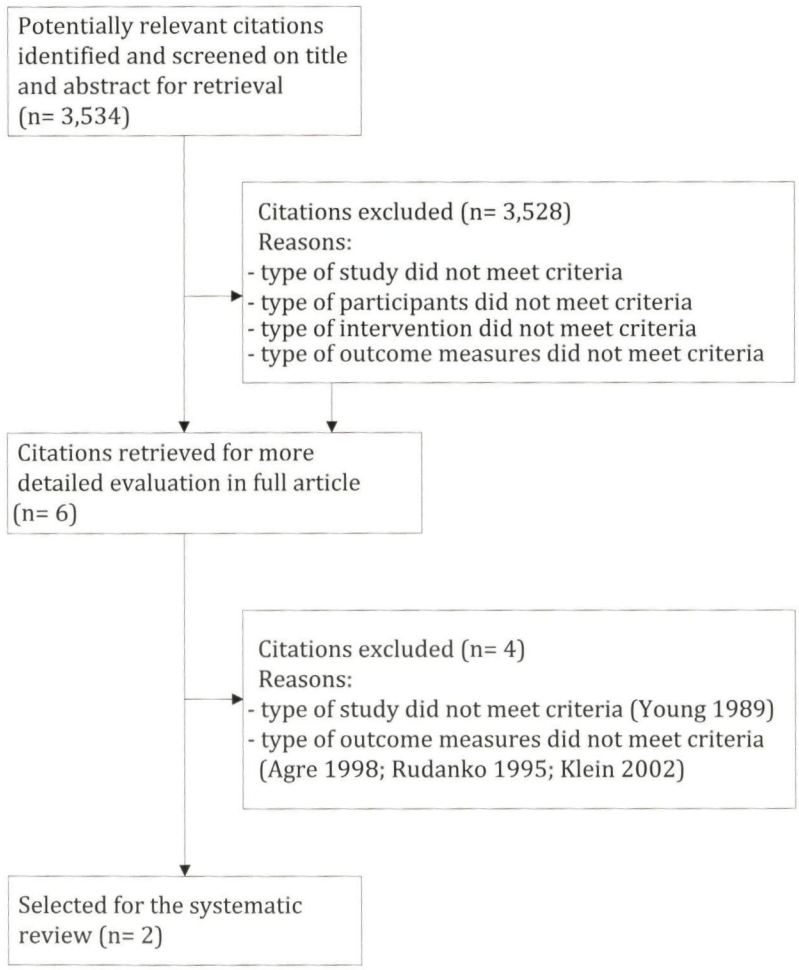

Figure 1. Flow chart showing the selection of studies for the systematic review.

Table 1. Criteria for methodological quality in the two studies selected for evaluation

Code	Description of the criteria	Aldehag et al. (2005)	Sandin-Aldehag & Jonsson (2003)
a	Were the eligibility criteria specified?	Yes	Yes
b	1) Was a method of randomization performed?	NA	NA
	2) Was the treatment allocation concealed?	NA	NA
c	Were the groups similar at baseline regarding the most important prognostic indicators?	NA	NA
d	Were the (index and control) interventions explicitly described?	Yes	Yes
f	Were co-interventions avoided or comparable?	Yes	Unknown
g	Was the compliance acceptable (in all groups)?	Yes	Yes
i	Was the outcome assessor blinded?	Unknown	Yes
j	Were the outcome measures relevant?	Yes	Yes
k	Were adverse effects described?	Yes	No
l	Was the withdrawal/drop-out rate described and acceptable?	Yes	Yes
m	1) Was a short-term follow-up measurement performed?	Yes	Yes
	2) Was a long-term follow-up measurement performed?	No	No
n	Was the timing the outcome assessment in all patients comparable?	Yes	Yes
o	Was the sample size for each group described?	Yes	Yes
p	Did the analysis include an intention-to-treat analysis?	Yes	No
q	Were point estimates and measures of variability presented?	Yes	Yes
Score for internal validity (criteria b,f,g,i,j,l,n,p)		6 of 9	5 of 9
Score for descriptive quality (criteria a,c,d,k,m)		4 of 6	3 of 6
Score for statistical quality (criteria o,q)		2 of 2	2 of 2

NA: Not Applicable.

Four of the six studies were excluded because either the type of study[9] or the type of outcome measures did not meet the predefined criteria[4;6;7] (Figure 1). The article by Young (1989) reviewed the late effects of poliomyelitis and the abstract referred to guidelines for occupational therapy assessment and treatment[7]. The full-text article showed that these guidelines were based on the author's experiences as a clinician and a polio survivor and on the literature. The reference list was reviewed but comprised no occupational

therapy intervention studies. Table 2 presents characteristics of the other three studies that were excluded from further analysis.

Two articles fulfilled all inclusion criteria and were subjected to the methodological assessment[5;8]. Both studies were both pre-post designs. The methodological quality of these studies was sufficient (Table 1). The study characteristics are described in Table 3. Both studies evaluated an individualized hand training program with silicone-based putty in five patients with myotonic dystrophy type 1[5] and 12 patients with Welander distal myopathy[8], respectively.

The first study resulted in improvements in self-rated performance and satisfaction with performance using the Canadian Occupational Performance Measure[5]. The second study showed significant improvements in activities of daily living[8]. Muscle strength improved significantly in both studies, grip force did not and the results on pinch grip were inconsistent. In the study by Aldehag et al. (2005), fine motor skills were assessed and showed significant improvements[5]. In the study by Sandin-Aldehag & Jonsson (2003), range of motion was measured and showed significant improvements in the extension of some finger joints[8].

Applying the best evidence synthesis to these data, we conclude that these studies provide some indications for the efficacy of a hand training program in muscle diseases (myotonic dystrophy and Welander distal myopathy) with initial scores of at least 3 out of 5 on manual muscle test in wrist and hand.

Discussion

Methods

In the current review, we choose to conduct a comprehensive search in a variety of large databases. We did not limit our search to randomized controlled trials and controlled clinical trials. We included other designs because occupational therapy is a relatively young profession with an even younger research tradition and literature about the efficacy of occupational therapy was expected to be sparse. Also, other research designs were included because neuromuscular diseases generally have a low prevalence and incidence, making it difficult to use research designs with comparisons of more than one group. We considered searching specific databases such as OT seeker and PEDro, but these databases are restricted to clinical trials and systematic reviews and draw their articles from the other databases already searched in this study.

Table 2. Characteristics of excluded studies

Author, date	Type of study	Type of participants	Type of intervention	Type of outcome measures
Agre et al. (1989)	Retrospective study with charts reviewed; a selection of the patients was evaluated at a follow-up visit	84 patients visiting a post-polio clinic, 79 met the criteria for poliomyelitis	Recommendations including: pacing, energy conservation, gentle stretching or strengthening exercise, use of orthotic devices, weight loss or nutritional counseling, gentle aerobic exercise, use of a cane and use of adaptive equipment	Unknown
Klein, et al. (2002)	A randomized parallel group study	29 polio survivors (15 men, 14 women); inclusion criteria: bilateral knee-extensor and hip-extensor strength grade 3 – 5; > 30 days of shoulder pain with daily activity	Group 1: home exercise program focused on strengthening and stretching the hip-extensor and knee-extensor muscles; Group 2: lifestyle modification techniques to avoid shoulder overuse; Group 3: both interventions	Pain (Visual Analogue Scales) during 4 resistance tests and after performing 3 simulated tasks; isometric knee-extension and hip-extension (hand-held dynamometer)
Rudanko, 1995	Uncontrolled clinical study	20 persons with Leber's hereditary optic neuropathy	Prescription of a wide variety of low vision aids	Type and frequency of prescription of low vision aids with correlation to World Health Organization categories of visual impairment

Table 3. Characteristics of included studies

Characteristic	Aldehag et al. (2005)[5]	Sandin-Aldehag & Jonsson (2003)[8]
No. of patients	5	12
Patient characteristics		
Inclusion criteria	Myotonic dystrophy with score of at least 3 of 5 on manual muscle testing in wrist and hand	Welander distal myopathy with score of 2 or 3 of 5 on manual muscle testing in extension of the fingers
Male/Female	2/3	5/7
Mean age (Y)	52.4 (range: 28 to 67)	62.5 (range: 39 to 83)
Outcome measures		
Primary	Performance and satisfaction with performance of problems in daily activities (COPM)	Activities of daily living (interview with ADL- Taxonomy); Life satisfaction (modified Life Satisfaction Checklist)
Secondary	1) Muscle strength of extensors and flexors of wrist and fingers (hand-held myometer); 2) Grip force and pinch grip (Grippit); 3) Fine motor control (Purdue Pegboard)	1) Muscle strength in extension of the fingers (Manual muscle testing); 2) Grip strength and pinch grip (peak, mean and 10-second value) (Grippit); 3) Range of Motion (goniometer)
Intervention		
Intensity	12 weeks, 3 times per week, 45 minutes	12 weeks, 3 times per week, 45 minutes
Content	Hand exercise program with isolated and mass movements with silicone-based putty and a stretching program	Hand exercise program with isolated and mass movements with silicone-based putty and a stretching program
Results		
Primary	Mean performance increased with 1.7 (range: 0.4-3.8); Mean satisfaction increased with 2.7 (range: 0-6.3)	ADL improved ($p = .01$) Changes in life satisfaction were not significant
Secondary	1) Muscle strength increased ($p < .05$); 2) Grip force and pinch grip did not change ($p > .05$); 3) Fine motor control improved ($p < .005$)	1) Muscle strength increased in left hand ($p = .01$); 2) No values of grip strength changed significantly; 3) For pinch grip, peak value and left hand 10-second value increased ($p = .04$); 4) Range of Motion improved

COPM = Canadian Occupational Performance Measure; ADL = Activities of Daily Living.

Of the 3,534 citations screened, only six studies were retrieved for detailed evaluation. The reason our search yielded so many citations that did not fulfill the predefined inclusion criteria was due to our comprehensive search strategy. Because we included many different study types, the search led to citations that included cohort studies on incidence or prevalence of symptoms or studies on measurement instruments. We used MeSH (including all subheadings) and free text words for different types of neuromuscular diseases, which resulted in citations with a broad a range of neuromuscular conditions including stroke and Parkinson's diseases and musculoskeletal problems, and studies with children, which we did not want to include.

Our search also included broad terms such as 'rehabilitation' and 'activities of daily living' so studies about occupational therapy not specifically indexed as such were not missed. This resulted in many citations on interventions other than occupational therapy. However, if we had narrowed the search terms to occupational therapy only, it would not have resulted in the articles retrieved.

Main results

The purpose of this study was to summarize the evidence for occupational therapy in patients with neuromuscular diseases. Based on two other research designs, we found indicative findings for the effectiveness of a hand training program to improve daily activities in patients with muscle disorders (Myotonic dystrophy and Welander distal myopathy). These studies also suggest that training of hand function may be efficacious in improving hand strength and fine motor skills[5;8]. No evidence was found for any other occupational therapy intervention in neuromuscular diseases.

Recommendations and conclusion

Based on our results and clinical experience, we have made recommendations for the type of studies, participants, interventions and outcome measures in future research.

The preferred types of studies are randomized controlled trials and controlled clinical trials. However, pre-post designs should be considered as acceptable alternatives, because of the limited number of eligible patients with the same neuromuscular diagnosis. Moreover, because there is little evidence available on occupational therapy in neuromuscular diseases, we recommend that future research (reviews and clinical studies) not be

confined to quantitative intervention studies. Descriptive studies and qualitative research may generate valuable information for occupational therapy on patient's needs and their adaptive strategies to manage their daily occupations and fulfill their life roles, such as the studies by Jönsson et al.[10] or Thorén-Jönsson & Grimby[11].

Regarding the type of participants, there are indications that different factors affect the process of adaptation in patients with different neuromuscular diseases. For instance, patients with facioscapulohumeral muscular dystrophy and hereditary motor and sensory neuropathy may need a different approach to reduce fatigue than patients with myotonic dystrophy[12]. Therefore, homogenous patient samples should preferably be included in future occupational therapy research.

Because the goal of occupational therapy intervention is to maintain or increase participation in daily occupations and life roles, research should focus on the efficacy of interventions with this aim. In patients with neuromuscular diseases, such interventions would typically include training of daily activities, with or without the use of assistive devices, modifying the work and home environments, and educating about work simplification and energy conservation techniques.

Based on the findings from qualitative studies, we recommend a client-centered approach that includes the following aspects: educating the patient about the disease because education plays an important role in his or her understanding of the need to implement adaptive strategies[9;10;13]; evaluating the patient's perception of his or her life history, personal values, goals and problems[3;10] informing the patient about the adaptive strategies available; and identifying the patient's adaptive strategies, which can be used in occupational therapy to empower the patient to make changes in his or her occupational performance[10].

To measure the efficacy of occupational therapy, the outcome measures should be in agreement with the type of intervention. We recommend client-centered measurement instruments at the level of activities and participation, such as the Canadian Occupational Performance Measure[14] and the Impact on Participation and Autonomy questionnaire[15-17].

The low yield of our search of only two pre-post designs about hand training programs for patients with muscle disorders demonstrates the limited scientific evidence for occupational therapy in patients with neuromuscular diseases. Although clinically there is no debate, further

exploration and efficacy studies are needed to provide occupational therapy for patients with neuromuscular diseases with a firm scientific base.

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Appendix A
Search strategy for systematic reviews on the effectiveness of occupational therapy and physical therapy for persons with neuromuscular diseases.

Searches for controlled trials are according to the recommendations of the Cochrane Neuromuscular Disease Group. The search strategies for neuromuscular diseases, occupational therapy and physical therapy are the same in all databases.

OVID Medline
Search strategy for controlled trials (Cochrane Neuromuscular Disease Group):

1 randomized controlled trial.pt.
2 randomized controlled trials/
3 controlled clinical trial.pt.
4 controlled clinical trials/
5 random allocation/
6 double-blind method/
7 single-blind method/
8 clinical trial.pt.
9 exp clinical trials/
10 (clin$ adj25 trial$).tw.
11 ((singl$ or doubl$ or tripl$ or trebl$) adj25 (blind$ or mask$ or dummy)).tw.
12 placebos/
13 placebo$.tw.
14 random$.tw.
15 research design/
16 (clinical trial phase i or clinical trial phase ii or clinical trial phase iii or clinical trial phase iv.pt.
17 multicenter study.pt.
18 meta analysis.pt.
19 prospective studies/
20 intervention studies/
21 cross-over studies/
22 meta-analysis/
23 (meta?analys$ or systematic review$).tw.
24 control$.tw.
25 or/1-24
26 limit 25 to humans

Search strategy for neuromuscular diseases:

27 neuromuscular diseases/ or exp *motor neuron disease/ or exp *muscular atrophy, spinal/ or exp *muscular disorders, atrophic/ or exp *neuromuscular junction diseases/
28 muscular diseases/ or exp *arthrogryposis/ or exp *compartment syndromes/ or exp *eosinophilia-myalgia syndrome/ or exp *isaacs syndrome/ or exp *mitochondrial myopathies/ or exp *muscle cramp/ or

exp *muscle weakness/ or exp *muscular disorders, atrophic/ or exp *myopathies, structural, congenital/ or exp *myositis/ or exp *myotonic disorders/ or exp *paralyses, familial periodic/ or exp *rhabdomyolysis/
29 peripheral nervous system diseases/ or exp *amyloid neuropathies/ or exp *brachial plexus neuropathies/ or exp *diabetic neuropathies/ or exp *guillain-barre syndrome/ or exp *isaacs syndrome/ or exp *mononeuropathies/ or exp *nerve compression syndromes/ or exp *neuralgia/ or exp *neuritis/ or exp *peripheral nervous system neoplasms/ or exp *polyneuropathies/
30 "hereditary motor and sensory neuropathies"/ or exp *charcot-marie-tooth disease/ or exp *refsum disease/
31 exp *Postpoliomyelitis Syndrome/
32 (neuromuscular disorder$ or neuromuscular disease$).mp. [mp=ti, ot, ab, nm, hw]
33 (muscular disease$ or muscular disorder$).mp. [mp=ti, ot, ab, nm, hw]
34 (muscle disease$ or muscle disorder$).mp. [mp=ti, ot, ab, nm, hw]
35 (neuromuscular junction diseases$ or neuromuscular junction disorder$).mp. [mp=ti, ot, ab, nm, hw]
36 (motor neuron disease$ or motor neuron disorder$).mp. [mp=ti, ot, ab, nm, hw]
37 (peripheral nervous system disease$ or peripheral nervous system disorder$).mp. [mp=ti, ot, ab, nm, hw]
38 (polyneuropath$ or neuritis or neuralgia).mp. [mp=ti, ot, ab, nm, hw]
39 (myopath$ or myotoni$).mp. [mp=ti, ot, ab, nm, hw]
40 myositis.mp. [mp=ti, ot, ab, nm, hw]
41 muscular dystroph$.mp. [mp=ti, ot, ab, nm, hw]
42 or/27-41

Search strategy for occupational therapy:
43 "activities of daily living"/ or occupational therapy/ or rehabilitation, vocational/
44 "activities of daily living"/ or automobile driving/ or exercise/ or leisure activities/ or work/
45 self-help devices/ or communication aids for disabled/ or wheelchairs/
46 Splints/
47 Counseling/
48 Caregivers/
49 assistive devices.mp. [mp=ti, ot, ab, nm, hw]
50 Splint$.mp. [mp=ti, ot, ab, nm, hw]
51 energy conservation.mp. [mp=ti, ot, ab, nm, hw]
52 carer$.mp. [mp=ti, ot, ab, nm, hw]
53 Counsel?ing.mp. [mp=ti, ot, ab, nm, hw]
54 Occupation$.mp. [mp=ti, ot, ab, nm, hw]
55 or/43-54

Search strategy for physical therapy:

56 behavior control/ or clinical protocols/ or drainage/ or exercise movement techniques/ or musculoskeletal manipulations/ or exp *physical therapy techniques/
57 exp *"Physical Therapy (Specialty)"/
58 rehabilitation/ or "activities of daily living"/ or early ambulation/ or physical therapy techniques/ or respiratory therapy/ or self care/
59 breathing exercises/ or relaxation techniques/
60 exercise movement techniques/ or breathing exercises/ or drainage, postural/ or exercise/ or exercise therapy/ or relaxation/ or relaxation techniques/
61 exercise/ or physical fitness/
62 exercise therapy/ or motion therapy, continuous passive/ or kinesiology, applied/ or manipulation, spinal/ or massage/
63 exercise movement techniques/ or breathing exercises/ or drainage, postural/ or exercise/ or exercise therapy/ or relaxation/ or relaxation techniques/ or walking/
64 hand strength/ or muscle fatigue/ or muscle tonus/ or musculoskeletal equilibrium/ or physical endurance/ or anaerobic threshold/ or exercise tolerance/ or posture/ or "range of motion, articular"/
65 "Physical Education and Training"/
66 (physiotherapy or physical therapy).mp. [mp=ti, ot, ab, nm, hw]
67 (exercise therap$ or exercise training or exercise program$ or training program$).mp. [mp=ti, ot, ab, nm, hw]
68 (strength training or aerobic training or aerobic exercise$).mp. [mp=ti, ot, ab, nm, hw]
69 (resistive exercise$ or endurance training or muscle exercise$).mp. [mp=ti, ot, ab, nm, hw]
70 or/56-69

OVID CINAHL

Search strategy for controlled trials (Cochrane Neuromuscular Disease Group):

1 random assignment/ or random sample/ or simple random sample/ or stratified random sample/ or systematic random sample/
2 Crossover design/
3 exp Clinical trials/
4 Double-blind studies/ or triple blind studies/
5 Placebos/
6 Quasi-experimental studies/
7 Solomon four-group design/ or Static group comparison/
8 Meta analysis/
9 Concurrent prospective studies/ or Prospective studies/
10 Factorial design/
11 ("clinical trial" or "systematic review").pt.
12 random$.tw.
13 ((singl$ or doubl$ or tripl$ or trebl$) adj25 (blind$ or mask$)).tw.
14 (cross?over or placebo$ or control$ or factorial or sham? or dummy).tw.

15 ((clin$ or intervention$ or compar$ or experiment$ or preventive or therapeutic) adj10 trial$).tw.
16 ABAB design$.tw.
17 (meta?analys$ or systematic review$).tw.
18 or/1-17
19 18 and subject terms

OVID Embase

Search strategy for controlled trials (Cochrane Neuromuscular Disease Group):
1 randomized controlled trial/
2 clinical trial/
3 multicenter study/
4 controlled study/
5 crossover procedure/
6 double blind procedure/
7 single blind procedure/
8 randomization/
9 major clinical study/
10 placebo/
11 meta analysis/
12 phase 2 clinical trial/ or phase 3 clinical trial/ or phase 4 clinical trial/)
13 (clin$ adj25 trial$).tw.
14 ((singl$ or doubl$ or tripl$ or trebl$) adj25 (blind$ or mask$)).tw.
15 placebo$.tw.
16 random$.tw.
17 control$.tw.
18 (meta?analys$ or systematic review$).tw.
19 (cross?over or factorial or sham? or dummy).tw.
20 ABAB design$.tw.
21 or/1-20
22 human/
23 nonhuman/
24 22 or 23
25 21 not 24
26 21 and 22
27 25 or 26
28 27 and subject terms

Part II: Scientific perspective

Chapter 5

Exercise therapy and other types of physical therapy for patients with neuromuscular diseases: a systematic review

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Abstract

Objective: to summarize and critically appraise the available evidence on exercise therapy and other types of physical therapies for patients with neuromuscular diseases (NMD).

Data Sources: Cochrane Central Register of Controlled Trials and Cochrane Database of Systematic Reviews, Medline, CINAHL, Embase (Rehabilitation and Physical Medicine), and reference lists of reviews and articles.

Study Selection: Randomized clinical trials (RCTs), controlled clinical trials (CCTs) and other designs were included. Study participants had to have any of the following types of NMD: motor neuron diseases, disorders of the motor nerve roots or peripheral nerves, neuromuscular transmission disorders or muscle diseases. All types of exercise therapy and other physical therapy modalities were included. Outcome measures had to be at the level of body functions, activities, or participation according to the definitions of the International Classification of Functioning, Disability and Health (ICF).

Data Extraction: Two reviewers independently decided on inclusion or exclusion of articles and rated the methodologic quality of the studies included. All RCTs, CCTs and other designs only if of sufficient methodological quality were included in a best evidence synthesis. A level of evidence was attributed for each subgroup of NMD and each type of intervention.

Data Synthesis: Initially 58 studies were included: 12 RCTs, 5 CCTs and 41 other designs. After methodologic assessment, 19 other designs were excluded from further analysis. There is level II evidence ("likely to be effective") for strengthening exercises in combination with aerobic exercises for patients with muscle disorders. Level III evidence ("indications of effectiveness") was found for aerobic exercises for patients with muscle disorders and for the combination of muscle strengthening and aerobic exercises in a heterogeneous group of muscle disorders. Finally, there is level III evidence for breathing exercises in patients with myasthenia gravis and for patients with myotonic muscular dystrophy. Adverse effects of exercise therapy were negligible.

Conclusions: the available evidence is limited, but relevant for clinicians. Future studies should be preferably multicentered, and use an international classification of the variables of exercise therapy and an ICF core set for NMD in order to improve comparability of results.

Introduction

Neuromuscular diseases (NMD) represent a heterogeneous group of disorders, including motoneuron diseases, disorders of motor nerve roots or peripheral nerves, neuromuscular transmission disorders and muscle diseases.[1;2] The progression of the diseases varies considerably. The deficits can range from muscle weakness, sensory loss, pain, fatigue and autonomic dysfunction in varying combinations. These deficits combine to lead to impairments of musculoskeletal and sensory functions, limitations in activities and restrictions in participation.[2]

There are approximately 600 different NMD with great variety in referral to physical therapy (PT).[3] There is no consensus regarding the type and intensity of PT.[4] PT often includes exercise therapy to improve or preserve muscle function (strength and endurance) and aerobic capacity to prevent or reduce secondary problems such as contractures, pain or fatigue.[2;4] The benefits or injurious effects of exercises in NMD are unclear, and this uncertainty is particularly pertinent when the consideration of the 'appropriate' level of intensity is addressed.[5-11]

Three Cochrane systematic reviews thus far focused on one type of NMD or a specific type of exercise therapy and were restricted to randomized clinical trials (RCTs) or controlled clinical trials (CCTs). In their Cochrane review on muscle strength training and aerobic exercise training for patients with muscle diseases, van der Kooi et al[12] concluded that moderate-intensity strength training in myotonic dystrophy and facioscapulohumeral muscular dystrophy appeared not to be harmful, but there was insufficient evidence to establish its benefit. This conclusion was based on 2 RCTs.[13;14] White et al.[15] did a Cochrane review on exercise therapy for people with peripheral neuropathy and concluded that there is insufficient evidence to evaluate the effect of exercise therapy on functional ability in people with this condition. This was based on 1 RCT,[13] which was also included in the review by van der Kooi.[12]. Ashworth et al, [16] reviewing treatment for spasticity in patients with amyotrophic lateral sclerosis (ALS), concluded that the available evidence was insufficient to determine whether individualized exercises for muscle endurance of the trunk and limbs with moderate intensity are beneficial or harmful for these patients. The conclusion was also based on 1 single RCT performed.[17]

When RCTs are scarce, evidence from nonrandomized studies and other designs, such as pre-post studies or case-control studies may be particularly relevant.[18] There are previous reviews on exercise therapy for

patients with NMD,[9;10] including other designs. However, these reviews lack a systematic approach.

Our aim here is to summarize and critically appraise the available evidence on exercise therapy and other modalities of physical therapies for patients with NMD to support neurologists, physicians, and physical therapists in their clinical decision-making for the individual patient with NMD. We conducted a comprehensive systematic review, including RCTs, CCTs, and other designs, all types of exercise and PT and all types of NMD.

Methods

Search strategy

We searched in the Cochrane Central Register of Controlled Trials (The Cochrane Library 2005, Issue 3), Ovid Medline In-Process & Other Non-Indexed Citations and Ovid Medline (1966 through September 2005), CINAHL WebSPIRS 5.1 (1982 through September 2005) and Embase WebSPIRS 5.03 (Rehabilitation and Physical Medicine) (1995 through September 2005). We also searched in the Cochrane Database of Systematic Reviews to find articles indirectly by screening reference lists. Further, potentially relevant publications were searched manually through citation and author tracking.

Our broad search strategy was built on the following components: 1) RCTs and CCTs as recommended by the Cochrane Neuromuscular Disease Group; 2) other designs, such as pre-post designs; 3) types of NMD; and 4) PT modalities. For all search strategies Medical Subject Headings (MeSH) or indexed terms were used as well as free-text words. The full search strategy can be found in Appendix A at the end of *Chapter 4*.

Selection criteria

Inclusion was restricted to articles with an abstract, and language had to be English, German, French, or Dutch. The following study designs were included: RCTs, CCTs and other designs such as pre-post studies. Case studies were excluded.

Participants included adults having one of the following types of NMD: 1) motoneuron diseases; 2) motor nerve root and peripheral nerve disorders; 3) neuromuscular transmission disorders; or 4) muscle disorders. Excluded were children or adolescents (< 18 y) with Duchenne's muscular dystrophy, spinal muscular atrophy or Becker's muscular dystrophy. Also, patients having signs and symptoms of muscle weakness, pain or fatigue, not related

to specific NMD were excluded as well as diagnoses including chronic fatigue syndrome, spinal cord injuries, thoracic outlet syndrome, reflex sympathetic dystrophy or complex regional pain syndrome, cancer or acquired immune deficiency syndrome. Although often classified as NMD, diabetic neuropathies and entrapment neuropathies such as carpal tunnel syndrome and radiculopathies were not included in this review.

Regarding the type of interventions, we included different modalities of PT: 1) muscle strengthening exercises; 2) aerobic exercises; 3) breathing exercises; 4) other interventions such as relaxation techniques, exercises to improve mobility including transfers and walking, functional electric stimulation, education of the patient, family and caregivers; or 5) a combination of these interventions. Comprehensive rehabilitation programs were excluded, since it is impossible to isolate the effectiveness of PT in such programs.

Outcome measures had to be at the level of body functions, activities or participation according to the definitions of the International Classification of Functioning, Disability and Health (ICF)[19]. Outcome measures at the level of body functions included measures for muscle strength or muscle endurance, range of motion, aerobic capacity, pulmonary function, respiration, pain, or fatigue. Excluded were measures for blood pressure as well as blood parameters, such as lactate. Outcome measures at the level of activities and participation included indices for walking and moving around, self care, work and employment, domestic life, leisure, quality of life, or general health.

Procedure for inclusion

In all databases a preliminary selection, based on title and abstract, was carried out by 2 reviewers (EHC and AJP) independently. Studies that seemed to fit inclusion criteria were retrieved for more detailed evaluation (Figure 1). In case of doubt, the reviewers collaboratively decided on retrieval of the full article.

Another pair of reviewers (EHC and JMB-P) independently decided on the inclusion or exclusion based on detailed information in the full articles. If disagreements persisted, a third reviewer (AJP) was consulted. The 2 reviewers (EHC and JMB-P) also independently scanned the reference lists of all articles and reviews for additional articles. Additional articles fulfilling the inclusion criteria, as well as relevant related articles and reviews found during the process of hand searching were retrieved for more detailed

evaluation. Final in- or exclusion of the articles was always based on independent assessments of 2 reviewers (EHC and JMB-P).

Methodologic quality

The methodologic quality of the RCTs, CCTs and other designs was rated using the list recommended by van Tulder et al.[20] The quality of the internal validity was scored with 11 criteria (random assignment, allocation concealed, care provider blinded, cointerventions standardized, compliance ≥ 70%, patient blinded, outcome assessor blinded, outcome measures relevant, drop-out rate acceptable, timing of outcome assessment comparable, intention-to-treat analysis). Descriptive quality was scored with 8 criteria (eligibility criteria, similarity of groups at baseline, interventions, adverse effects, short-term follow-up, long-term follow-up, sample size, point estimates and measures of variability). All criteria were scored as yes, no, or unclear. Equal weight was given to all items. For the other designs, only the criteria that were applicable (7 criteria for internal validity and 7 criteria for descriptive quality) were used.

RCTs and CCTs were considered to be of sufficient quality if at least 6 of 11 criteria for internal validity and 4 of 8 descriptive criteria were scored positively. The other designs were considered to be of sufficient quality if at least 4 of 7 criteria for internal validity and 4 out of 7 descriptive criteria were scored positively. Two reviewers (EHCC and JMCB-P) independently rated the methodological quality of the trials. Disagreements were resolved by discussion.

We decided to exclude other designs of insufficient methodologic quality from further analysis because of their lack of value for the best evidence synthesis.

Classification of included studies

The studies were classified based on type of NMD and type of intervention. For each study, the design, the methodologic quality, the number of participants, the diagnosis, the exercise intensity, frequency and duration, and outcome was presented (Table 1).

Studies comparing a group of patients with a reference group of healthy persons were categorized as other designs. Also studies in which 1 side of the body was exercised and the contralateral side of the body served as control were treated as other designs.

The type of intervention was categorized into the different modalities of PT: 1) muscle strengthening exercises; 2) aerobic exercises; 3) breathing exercises; 4) other interventions; or 5) a combination of these modalities. For the different types of exercise therapy, the intensity was described, if the article provided enough details.

We divided the outcome variables into 2 categories: 1 category with outcome variables at the level of body functions and a second category including outcome variables at the level of activities and participation. This is based on the corresponding components in the ICF.[19] Adverse effects were presented separately. The aim was to present primary and secondary outcomes for each study. However, only few studies defined primary and secondary outcome measures.[14;21] We then decided to include all outcome variables fulfilling the inclusion criteria and considered them of equal importance. Within each category (body functions or activities/participation respectively), the number of variables that showed a statistically significant ($P<.05$) effect in a study was divided by the total number of outcome variables in the study. If presented in the article, different subscales of an instrument were treated as different outcome variables. A study was considered to provide evidence of effectiveness if more than half of the variables showed a significant effect.

Best evidence synthesis

The comprehensive scope of our review and the heterogeneity in patient populations, interventions and outcome measures precludes quantitative analysis (meta-analysis) of the data. We used a best evidence synthesis based on a classification of the Dutch Institute for Healthcare Improvement.[22] The following levels of evidence were attributed based on the hierarchy of evidence. Recommendations were formulated accordingly.

Level I evidence is based on (a systematic review of) at least 2 independent RCTs of sufficient methodologic quality and leads to the conclusion: "It has been shown that...".

Level II evidence is based on 1 good quality RCT or at least 2 independent controlled studies (RCTs or CCTs) of less methodologic quality and leads to the conclusion: "It is likely that...". Level III evidence is based on an RCT or CCT of low methodologic quality or at least 1 other design of sufficient methodologic quality. The conclusion is formulated as "There are indications that....". In case of inconsistent findings in studies of similar design and methodologic quality (studies with and without evidence), the conclusion is

formulated as 'There is insufficient evidence that...'. If studies showed consistent significant findings, a level of evidence was attributed for each subgroup of NMD and each type of intervention.

Results

Selection of studies

The search resulted in 5712 citations (see Fig 1). Of these, 5634 articles were excluded because the type of study, the participants, the intervention, or outcome measures did not meet the predefined criteria.

The preliminary selection resulted in 62 articles and 16 reviews that were retrieved for more detailed evaluation. Citation tracking resulted in 19 additional reviews and 27 additional articles to be retrieved. The final selection resulted in a total of 60 articles, comprising 58 different studies, because 2 studies were published twice.[13;23-25] Among these studies were 12 RCTs, 5 CCTs, and 41 other designs.

Methodologic quality

Five of 12 RCTs[13;14;21;26;27] and 1 of 5 CCTs[28] had sufficient methodologic quality (a score of ≥ 6/ 11 for internal validity and ≥ 4 8 for descriptive quality). Of the other designs, 22 studies had sufficient methodologic quality (a score of ≥ 4/7 for internal validity and ≥ 4/7 for descriptive quality). Nineteen other designs had insufficient methodologic quality and were excluded from further analysis.[29-47]

Two RCTs[17;48] had a lower score on the validity criteria than we accepted from other designs. Four RCTs[48-51] and 4 CCTs[28;52-54] presented only within-groups analysis, but no between-groups comparisons. These studies were treated as other designs in the best evidence synthesis.

The methodologic quality of all studies included in the best evidence synthesis is presented in Table 1.

Best evidence synthesis

The best evidence synthesis is based on the information presented in Table 1. For each subgroup of NMD and each type of intervention, the evidence is summarized.

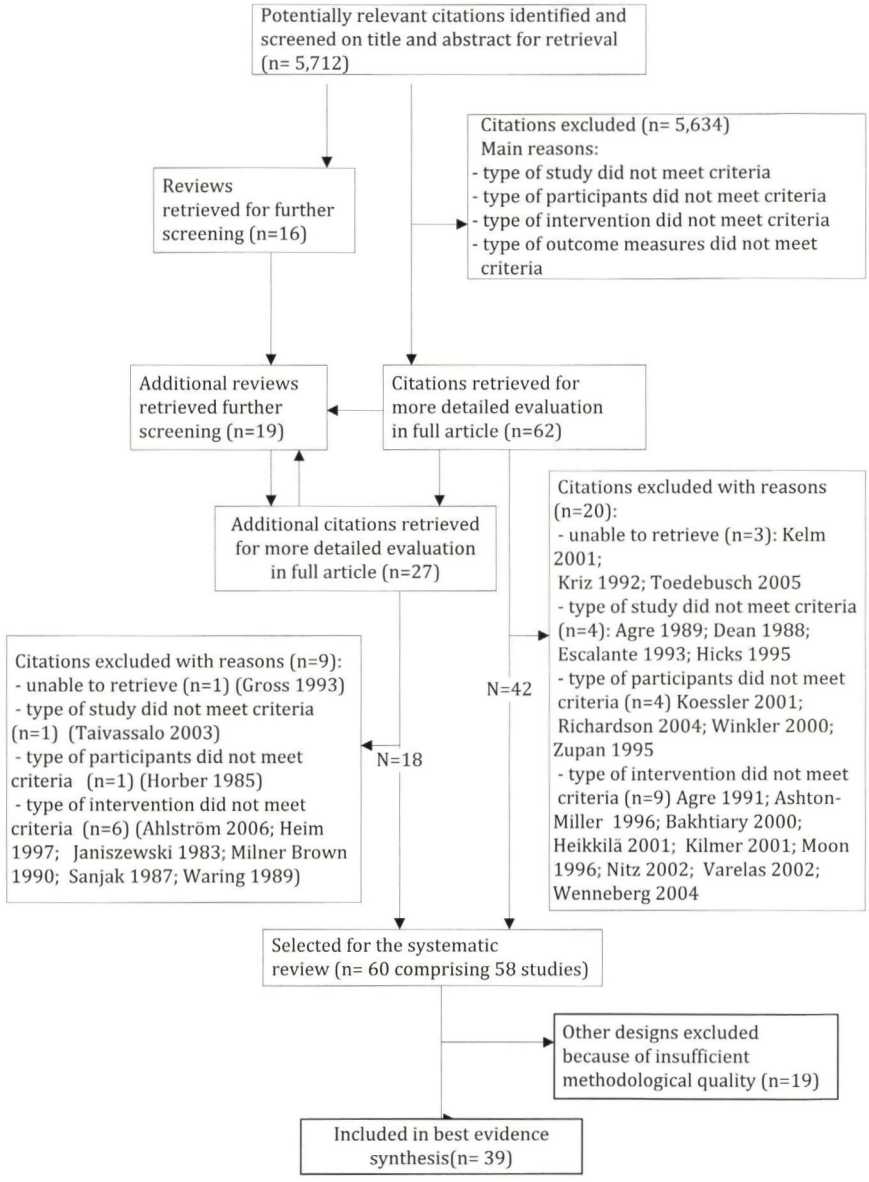

Figure 1. Flow chart showing the selection of studies for physical therapy for patients with neuromuscular disorders.

Motoneuron disorders

Muscle strengthening exercises. Four RCTs[17;48;50;55] and 4 other designs[56-59] studied muscle strengthening exercises in NMD. None of the RCTs had sufficient methodologic quality. One RCT[17] included patients with ALS and all other studies included patients with postpoliomyelitis syndrome (PPS). The low quality RCT[17] on muscle strengthening exercises in patients with ALS reported significant findings in 1 of 8 variables on body functions and 1 of 4 variables on activities and participation. According to the criteria used in this review, this implies that there is insufficient evidence for the effectiveness of muscle strengthening exercises for patients with ALS.

Three RCTs[48;50;55] of insufficient methodologic quality and 4 other designs[56-59] studied muscle strengthening exercises in PPS. The type and intensity of strengthening exercises and the outcome variables differed in all studies. Although 4 studies (1 RCT and 3 other designs) showed significant effects in most outcome variables at the level of body functions, the other 3 studies (2 RCTs and 1 other design) resulted in insufficient evidence of effectiveness. Hence, according to our criteria, there is insufficient evidence for the effectiveness of muscle strengthening exercises for patients with PPS.

Aerobic exercises. One RCT[60] and 1 CCT[53], both of limited methodologic quality, studied the effectiveness of aerobic exercise in PPS. The RCT showed a significant effect in 4 of 7 outcome variables in the category body functions. The CCT showed no significant results in the between-groups analysis. In the within-group analysis, significant results were found for 2 of 7 variables on body functions and in 1 of 2 variables on activities and participation level. Our conclusion is that there is insufficient evidence for the effectiveness of aerobic exercises for patients with PPS.

A combination of muscle strengthening and aerobic exercises. One CCT[61] of insufficient methodologic quality evaluated the combination of muscle strengthening exercises and aerobic exercises in patients with PPS. Three out of 20 variables on body functions and none of the outcome variables on activities and participation showed a significant effect. Hence, there is insufficient evidence for the effectiveness of a combination of muscle strengthening exercises and aerobic exercises for patients with PPS.

Lifestyle modification with or without muscle strengthening exercises. One study with a randomized parallel group design evaluated the effectiveness of lifestyle modification alone and in combination with muscle strengthening exercise in PPS[50]. The methodologic quality of the study was insufficient. There was a significant effect in 1 of 5 variables on body

functions in the group receiving the lifestyle modification. The group combining the 2 intervention strategies showed no significant findings. In conclusion, there is insufficient evidence for the effectiveness of lifestyle modification techniques, with or without muscle strengthening exercises for patients with PPS.

Motor nerve root disorders and peripheral nerve disorders

Muscle strengthening exercises. One RCT[13] of sufficient methodologic quality studied the effect of muscle strengthening exercises for patients with hereditary motor and sensory neuropathy (HMSN). Significant findings were found in 1 of 4 variables on body functions and 1 of 6 variables on activities and participation[13;25]. One other design[13;24] showed significant findings in 3 of 9 variables on body function and all 4 variables on activities and participation. Based on these findings, the conclusion is that there is insufficient evidence for the effectiveness of muscle strengthening exercises in patients with HMSN.

A combination of muscle strengthening and aerobic exercises. One RCT[51] of insufficient methodologic quality studied the effectiveness of muscle strengthening and aerobic exercises in chronic peripheral neuropathy (CPN). There were significant findings in 2 of 4 variables on body functions and in 1 of 11 variables on activities and participation. Our conclusion is that there is insufficient evidence for the effectiveness of strengthening exercises in combination with aerobic exercises in patients with CPN.

Neuromuscular transmission disorders

Breathing exercises. One RCT with sufficient methodologic quality studied the effectiveness of inspiratory muscle training and diaphragmatic breathing and pursed lips breathing in patients with myasthenia gravis (MG) [26]. Four of 19 variables on body functions and 1 of 9 variables on activities and participation showed a significant effect in the analysis between groups. The within-groups analysis showed a significant effect in 10 of 19 variables on body function. If we consider this study as an other design, then the conclusion is that there are indications for the effectiveness of breathing exercises in patients with MG (level III evidence).

Table 1. Classification of PT studies in type of NMD

Study	Design	V	D	N	Diagnosis	Type of intervention (type of exercises, type of muscles, load, number of reps, sets and progression)	Frequency and duration	Body functions	Activities/ participation
1. Motoneuron disorders									
Muscle strengthening exercises									
Drory et al[17]	RCT†	3	7	25	ALS	AROM exercises for muscles of limbs and trunk against modest loads	Twice a day 15 min for 12 mo	1/8†	1/4†
Klein et al[50]	RCT*	4	4	29	PPS	3-5 AROM exercises for hip and knee extensors against gravity. RPE 12-14, increasing number of reps	Daily 30 min for 16 wk	2/5*	NV
Prins et al [55]	RCT†	4	5	16	PPS	Aquatic exercise: swimming and exercises for arms and legs using fins and paddles	3 x/wk 45-70 min for 8 wk	7/39†	NV
Chan et al[48]	RCT*	3	7	10	PPS	3 sets of eight 3-5 seconds 50% MVC of thenar muscle. If possible increase of 10% per week until 70% MVC level	3 x/wk for 12 wk	*2/2*‖*	NV
Fillyaw et al[59]	OD‡	4	5	17	PPS	Full AROM exercises for knee or elbow: 3 sets of 10 reps with weights. 1st set weight of 50% of 10-RM; 2nd set 75% and 3rd set 100% of 10RM. 10RM weight was evaluated every 2 wk	Every other day up to 2 y	*2/3‖*	NV
Einarsson [58]	OD§	4	7	12	PPS	12 sets of 8 maximal isokinetic knee extensors contractions at 180°/sec angular speed interposed with 12 sets of isolated 4/s isometric contractions	3 x/wk (96 s) for 6 wk	*5/7‖*	NV
Agre et al[56]	OD	5	6	12	PPS	6 reps (30 s each) of knee extension with weight of 1-1,5 kg until RPE 17 or until 10 reps. If RPE <17 weight was increased (.23 kg)	Every other day up to 12 wk	0/5	NV
Agre et al[57]	OD	5	6	7	PPS	Tuesday and Friday : 3 sets of 12 reps with ankle weights (1–1,5 kg). If RPE <19, the weight increased (0.23 kg) the next session. Monday and and Thursday : 3 sets of 4 max effort isometric quadriceps contractions (5s)	4 d/wk for 12 wk	6/6‖	NV

Aerobic exercises									
Jones et al[60]	RCT†	5	7	37	PPS	Cycle ergometer at intensity of 70-75% of HRR plus resting HR. Bouts of 2-5 min, 1-min rest	3 x/wk 15-30 min for 16 wk	*4/7†*	NV
Dean and Ross[53]	CCT*†	4	6	20	PPS	Treadmill walking at comfortable speed, RPE ≤ 5 (somewhat heavy)	3 x/wk 20-40 min for 6 wk	*0/1† 2/7**	*1/2**
Muscle strengthening and aerobic exercises									
Willen et al[61]	CCT†	4	5	28	PPS	Fitness in water: resistance and endurance activities, balance, stretching and relaxation, avoiding mucle fatigue	2x/wk 40 min for 8 mo	*3/20†*	*0/4†*
Lifestyle modification									
Klein et al[50]	RCT*	4	4	29	PPS	Lifestyle modification to avoid shoulder overuse	Monthly for 16 wk	*1/5**	*NV*
Muscle strengthening exercise and lifestyle modification									
Klein et al[50]	RCT*	4	4	29	PPS	3 - 5 AROM exercises for hip and knee extensors against gravity, increasing number of reps. RPE 12-14	Daily 30 min for 16 wk	*0/5**	*NV*
2. Motor nerve root disorders and peripheral nerve disorders									
Muscle strengthening exercises									
Lindeman et al[13;25]	RCT†	6	7	29	HMSN	Exercises of muscles of hip and knee. Wk 1-8: 3 sets of 25 reps of 60% of 1-RM; wks 9-16: 3 sets of 15 reps of 70% of 1-RM; from wk 18: 10 repetitions of 80% of 1-RM	3 x/wk 30 min for 24 wk	1/4†	1/6†
Chetlin[23;24]	OD	5	6	20	HMSN	2 sets of varying reps for flexion and extension at 40% to 50% MVIS for knee and 20% to 30% MVIS for elbow. Every 4 wk, resistance increased. Reps increased wkly from 4 to 6 to 8 to 10	Daily for 12 wk	3/9	4/4‖
Muscle strengthening and aerobic exercises									

Ruhland and Shields[51]	RCT*	4	5	28	CPN	Exercises with Theraband with no resistance and progressing to light and medium resistance with 10 reps. Aerobic conditioning at 60-70% of estimated maximal HR or 'somewhat hard' on Borg RPE Scale	Daily 20 min for 6 wk	2/4*	1/11*
3. Neuromuscular transmission disorders									
Breathing exercises									
Fregonezi et al[26]	RCT†*	6	6	27	MG	Ten minutes diaphragmatic breathing, followed by 10 min interval-based inspiratory muscle training and 10 min of pursed lips breathing. Initial load 20% of Pimax, increased to 30% in wk 3, 45% in the fifth wk and 60% in wk 7.	3 x/wk 45 min for 8 wk	4/19† *10/19*‖* /	1/9† 1/9*
4. Muscle disorders									
Muscle strengthening exercises									
Van der Kooi et al.[14]	RCT†	7	7	65	FSHD	Dynamic and isometric exercises for elbow flexors and ankle dorsiflexors, wk 1-8: 2 sets of 5-10 reps with 10-RM weights, interspersed with 30 sec isometric exercise with 10-RM; wk 9-17: sets of 8 reps with 8RM weights; from wk 18: 5 reps of 5-RM.	3 x/wk for 52 wk	2/12†	0/3†
Lindeman et al[13;25]	RCT†	6	7	33	MMD	Exercises of muscles of hip and knee. Wks 1-8: 3 sets of 25 reps of 60% of 1-RM; wks 9-16: 3 sets of 15 reps of 70% of 1-RM; From wk 18: 10 reps of 80% of 1-RM	3 x/wk 30 min for 24 wk	0/4†	0/6†
Belanger and Noel[52]	CCT*	4	6	6	MMD (5) LGD (1)	Maximal contractions of dorsiflexors and plantarflexors with ankle exerciser. Two sets of 10 MVC of 3-5 s for dorsiflexors and for plantarflexors	Daily for 4 mo	0/2*	0/4*
Alexander son et al[63]	OD	6	6	11	PM (7) DM (4)	Warming up. Exercises for shoulder mobility and grip strength with a pulley apparatus, exercises for quadriceps and hip muscles, sit ups and stretching. If FI score > 38 exercises with weights (0.25 -2 kg) were included. Also 15- min walking at self-selected speed	5 x/wk 15 min for 12 wk	*4/5‖*	*3/8*

Arnadottir et al[64]	OD	6	5	7	sIBM	Exercises for shoulder mobility with a pulley apparatus, resistive exercises for shoulder and hip muscles, quadriceps and neck and trunk muscles. (10 reps of each exercise); Weight cuffs (0.25- 2 kg) depending on FI score (> 38) and self paced walking (15 min)	5 x/wk for 12 wk	0/3	NV
Aldehag et al[62]	OD	6	6	5	MMD 1	Hand exercises with silicone-based putty with isolated (1-2-3 sets of 3 reps or 1-2-3 sets of 5 reps) and mass (1-2-3 sets of 10 reps or 1-2-3 sets of 15 reps) movements, starting with 1 set of 3-5 reps in isolated movements and 1 set of 10-15 reps in mass movements. Number of sets increased every 4 wks. Stretching was also included	3 x/wk 45 min for 12 wk	*7/12*[ll]	*3/4*[ll]
Sandin and Jonsson[65]	OD	5	5	12	WM	Hand exercises with silicone-based putty with isolated (1-2-3 sets of 3 reps or 1-2-3 sets of 5 reps) and mass (1-2-3 sets of 10 reps or 1-2-3 sets of 15 reps) movements, starting with 1 set of 3-5 reps in isolated movements and 1 set of 10-15 reps in mass movements. Number of sets increased every 4 wks. Stretching was also included	3 x/wk 45 min for 12 wk	*4/14*	*1/2*
Tollbäck[66]	OD‡	4	6	9	MMD	Maximal AROM exercises for knee extensors with weights on an iron shoe with increasing load. Wk 1: 60% of 1-RM; wk 2: 70% of 1-RM and thereafter 80% of 1- RM. 3 sets of 8 reps were performed	3 x/wk for 12 wk	1/4	NV
Aerobic exercises									
Taivassalo et al[70]	OD§	4	5	24	MM (14) NMM (10)	Treadmill exercise 70 to 85% of HRR	3-4 x/wk 20-30 min for 8 wk	2/2[ll]	1/1[ll]
Taivassalo et al[69]	OD	5	5	10	MM	Treadmill exercise at 60-80% of HRR Exercise until reaching the RPE of 15 ("hard")	3-4 x/wk 20-30 min for 8 wk	4/4[ll]	1/1[ll]

Taivassalo et al[71]	OD	5	5	10	MM	Cycling at 70-80% of maximal HR	3-4 x/wk 30-40 min for 14 wk	4/6‖	NV
Trenell et al[72]	OD§	5	5	10	MM	Cycling at 70-80% of maximal HR	3 x/wk 30 min for 12 wk	2/3	1/1
Haller et al[67]	OD	4	5	8	MD	Cycling at 60-70 % of maximal HR	4 x/wk 30-40 min for 14 wk	3/3	NV
Olsen et al[68]	OD	5	5	8	FSHD	Cycling at 65% of VO_2 max	5 x/wk 35 min for 12 wk	2/2‖	NV
Sunnerhagen et al[73]	OD	5	5	8	HM	Cycling at 70% of maximal workload Subjective workload with a target of 5 (Borg category scale with exponential increments from 0-10)	5 x/wk 30 min for 8 wk	1/3	1/4
Muscle strengthening and aerobic exercises									
Cejudo et al[49]	RCT*	4	7	20	MM	Cycling 30 min at 70% of peak work rate; and 3 dynamic isotonic arm weight (50% 1-RM, repeated every 2 wk) lifting procedures through full ROM; shoulder press (shoulder, upper back and arm muscles), butterfly (pectoralis major) and biceps curls (biceps brachii and brachialis). Wk 1 and 2: 1set of 10-15 reps; next wks: 2 -3 sets	3 x/wk 60 min for 12 wk	13/18*‖	*2/4*‖*
Wiesinger et al[27]	RCT†	6	7	14	DM/PM	Cycling: 3-5 min warming up; resistance increased until 60% of maximal HR. Step aerobics 30 min at different rate levels adjusting load of exercises. Last 5 min cooling down and stretching exercises. First 2 wks 2 times/wk, remaining 4 wks: 3 x/wk	2-3x/wk 60 min for 6 wk	2/2†‖	1/1†‖

Wiesinger et al[54]	CCT*	5	7	8	DM/PM	Cycling: 3-5 min warming up; resistance increased until 60% of maximal HR. Step aerobics 30 min at different rate levels adjusting load of exercises. Last 5min cooling down and stretching exercises. First 2 wk 2x/wk, 4 wk:3x/wk, remaining 18 wk: 1x/wk	1-3x/week 60 min for 6 mo	6/8*‖	1/1*‖
Muscle strengthening exercises, respiratory exercises and mud/massage/bath									
Varju et al[74]	OD	4	5	21	DM/PM	Assisted bending and stretching of all joints; Isotonic muscle training until fatigue. Movements were repeated until 65-70% of max reps, then they had rest for 3min. If muscle were too weak a sling was used. Breathing exercises included instructions how to put hands on abdomen and rib cage and feeling and controlling movements, mud/massage/bath	5 x/wk 40-60 min for 3 wk	9/18	2/4
Breathing exercises									
Ugalde et al[75]	OD§	4	5	11	MMD	Pursed lips breathing as voluntary expiratory blowing through partially closed lips to create a resistance at the mouth compared to matched volume breathing (exhaling without pursing lips) and tidal breathing (comfortable breathing without pursing lips)	120 s of each breathing condition	6/9‖	NV
5. Heterogeneous neuromuscular disorders									
Strengthening exercises									
Aitkens et al[76]	OD‡§	4	4	27	MMD (12); HMSN (8); LGD (3); SMA (2); FSHD (2)	Initially 3 sets of 4 reps with speed of 30/s for knee extensor with ankle weights of 30% of max isometric knee extension force; and also for elbow flexors with weight of 10% of max elbow flexion force. Also 3 sets of 4 reps for hand grip exercises. Increasing resistance and reps	3 x/wk 15-20 min for 12 wk	7/11‖	NV

Kilmer et al[77]	OD‡§	6	5	10	MMD (5); LGD (3); HMSN (2)	Knee extensors and elbow flexors with ankle and wrist cuff weights, 1 set of 10 reps with 12- RM, gradually increasing until 5 sets of 10 reps during 4 d/wk. After wk 5, resistance was increased by .45kg/wk (1lb/wk) if feasible. After wk 9, from 4 to 5 sets	3-4x/wk for 12 wk	3/10	NV
Aerobic exercises									
Florence And Hagberg[28]	CCT*	6	5	12	CCD (1); NM (1); MyM (1); SMA (1); CMD (2); LGD (3); CMT (2); FSHD (1)	Cycling on cycle ergometer 5-min exercise bouts, 2-min rest at 70% of VO_2max: Resistance was adjusted to continually elicit 70% of VO_2max	3 x/wk for 12 wk	3/5*‖	0/2*
Wright ett al[78]	OD	4	5	11	MMD (7); HMSN (3); LGD (1)	Walking with training at HR of 50-60% HRR Wk 1 and 2: 3 times per wk 15 min; week 3: 20-30 min; wk 5 – 9 options to increase frequency to 4 x/wk	3-4x/week 15-20-30min 12 wk	1/33	NV
Muscle strengthening and aerobic exercises									
Dawes et al[21]	RCT†*	7	6	18	Becker MD (4); MMD (4); PM (1); FSHD (3); IMB (1); LGD (4); CM (1)	Walking for as long as possible at light subjective exercise intensity up to 20 min, then increase towards a moderate intensity (Borg CR-10 scale). Two exercises for each leg muscle endurance and core stability increasing the number of reps and range until 2.5 min for each exercise. Then increasing difficulty with gravity as resistance, increasing number of reps	Alternate days for 8 wk	1/7† 6/7*‖	0/6† 0/6*

Abbreviations: Activities/participation = number of variables at the level of activities or participation with significant change divided by total number of variables at the level of activities or participation; ALS = amyotrophic lateral sclerosis; AROM = active range of motion; Becker MD = Becker muscular dystrophy; Body functions = number of variables at the level of body functions with a significant change divided by the total

number of variables at the level of body functions; Borg CR-10 scale = exercise symptom rating scale; CCD = central core disease; CM = Congenital myopathy; CMD = Congenital Muscular Dystrophy; CMT = Charcot-Marie-Tooth; CPN = Chronic Peripheral Neuropathy; D = descriptive criteria (maximum, 8); DM = dermatomyositis; Fi = functional index in myositis; FSHD = facioscapulohumeral muscular dystrophy; HM = hereditary myopathy; HMSN = hereditary motor and sensory neuropathy; HR = heart rate; HRR = heart rate reserve; IBM = inclusion body myositis; LGD = limb girdle dystrophy; MD = McArdle's disease; MG = myasthenia gravis; Min = minutes; MM = mitochondrial myopathy; MMD = myotonic muscular dystrophy; MVC = maximum voluntary contraction; MVIS = maximum voluntary isometric strength; MyM = myotubular myopathy; N = number of patients included in the study; NM = nemaline myopathy; NMM = nonmetabolic myopathy; NV = no variables in this area; OD = other design; PM = polymyositis; PPS = postpoliomyelitis syndrome; reps = repetitions; RPE = rating of perceived exertion; s = seconds; sIBM = sporadic inclusion body myositis; SMA = spinal muscular atrophy; V = criteria for internal validity (maximum, 11); VO_2max = maximum oxygen uptake; wk = week; WM = Welander myopathy; xRM = repeated maximum (maximum weight, which can be lifted x repetitions).

*Analysis within groups;

† Analysis between groups;

‡ One side of the body randomly chosen for exercise, contralateral side of the body served as control;

§ Comparison with a reference group of healthy subjects;

‖ Results significant (P<.05) for more than half of the variables.

Muscle disorders

Muscle strengthening exercises. Two RCTs[13;14;25] of sufficient methodological quality, one CCT[52] with insufficient methodologic quality and 5 other designs[62-66] studied strengthening exercises for patients with muscle disorders. Different types of muscle disorders and different muscle groups were studied. There was also much variety in type and intensity of strengthening exercises and in the outcome variables. The RCTs and CCT showed hardly significant findings, neither at the level of body functions nor at the level of activities and participation. The findings of the other designs were inconsistent. In conclusion, there is insufficient evidence for the effectiveness of muscle strengthening exercises for patients with muscle disorders.

Aerobic exercises. Seven other designs[67-73] studied aerobic exercises for patients with muscle disorders. Six other designs [67-72] showed consistent significant findings at the level of body functions and at the level of activities and participation. One other design[73] showed significant findings in only 1 of 3 outcome variables on body function. The outcome variable that showed a significant effect was a measure of aerobic capacity; the other 2 were strength measures. In conclusion, there are indications that aerobic exercises have a positive effect on body functions as well as activities and participation in patients with muscle disorders (level III evidence).

A combination of muscle strengthening and aerobic exercises. Two RCTs[27;49] and 1 CCT[54] studied the combination of muscle strengthening exercises and aerobic exercises in patients with muscle disorders. One RCT[27] had sufficient methodologic quality, whereas the methodologic quality of the other RCT[49] and the CCT[54] was insufficient. The good quality RCT[27] showed significant findings in all 2 variables at the level of body functions and in the only variable measured at the level of activities and participation. The low quality RCT[49] and CCT[54] showed significant findings consistent with the good quality RCT. In conclusion, it is likely that strengthening exercises in combination with aerobic exercises have a positive effect on body functions as well as on activities and participation in patients with muscle disorders (level II evidence).

A combination of muscle strengthening and breathing exercises and mud/massage/bath. One other design[74] showed significant findings in 9 of 18 variables at the level of body functions and 2 of 4 variables at the level of activities and participation. In conclusion, there is insufficient evidence for

the effectiveness of the combination of strengthening and respiratory exercises and mud/massage/bath for patients with muscle disorders.

Breathing exercises. One other design[75] on pursed lips breathing in myotonic muscular dystrophy (MMD) showed significant findings on 6 of 9 variables on body functions. In conclusion: there are indications that pursed lips breathing is effective patients with MMD (level III evidence).

Heterogeneous group of patients with NMD

Muscle strengthening exercises. Two other designs[76;77] studied muscle strengthening exercises in a heterogeneous group of patients with NMD. The findings were inconsistent. In conclusion, there is insufficient evidence for the effectiveness of muscle strengthening exercises for a heterogeneous group of NMD.

Aerobic exercises. One CCT[28] and other design[78] studied aerobic exercises in a heterogeneous group of patients with NMD. The CCT was of sufficient methodologic quality and presented significant findings in 3 of 5 variables on body function[28]. There were no significant findings at the level of activities and participation. Only within-groups findings were presented, which means that we cannot consider this study to be a truly controlled trial. The other design[78] showed a significant effect in only 1 of 33 variables on body functions. In conclusion, there is insufficient evidence for the effectiveness of aerobic exercises for a heterogeneous group of NMD.

Muscle strengthening and aerobic exercises. One RCT[21] of sufficient methodologic quality studied muscle strengthening and aerobic exercises in a heterogeneous group of NMD. The between-groups analysis showed a significant effect in 1of 7 variables on body functions and none of the variables at the level of activities and participation. The within-group analysis showed a significant effect in 6 of 7 strength measures in the intervention group. If we consider this study as an other design, then the best evidence synthesis leads to the conclusion that there are indications that a combination of muscle strengthening and aerobic exercises is effective in increasing muscle strength (level III evidence).

Adverse effects

Thirty-three studies reported absent or negligible adverse effects. Chetlin et al.[24] reported that 3 of 20 patients decreased their training for 1 or 2 sessions due to delayed-onset soreness. Six studies did not report whether exercise therapy resulted in adverse effects.

Other PT modalities

No studies were found on the application of functional electrical stimulation or interventions to improve mobility including transfers and walking or education of the patient, family and caregivers.

Discussion

Methodology

The extensive search used in this review with MeSH terms exploded without restrictions and free text words such as activities of daily living or physical activity, resulted in a large database of citations. However, 90% of the citations did not fulfill our predefined inclusion criteria regarding the study design, the participants, the intervention, or the outcome measures. Surprisingly, hand searching of the reference lists of the articles and reviews revealed a substantial number of additional citations. Hopewell et al.[79] compared hand searches versus Medline searching and found that 25 % of reports of randomized trials with a Medline record were missed by the electronic search, because they did not have either of the publication type terms *randomized controlled trial* or *controlled clinical trial.* This was especially the case for reports of RCTs published prior to 1991. It shows the limitations of electronic searches and suggests the need to combine electronic searches and manual searches.[18] Another explanation is that the indexing of other designs is less precise and reliable compared with RCTs.[18]

We included all RCTs and CCTs in our best evidence synthesis, regardless of their methodologic quality and only the other designs of sufficient methodologic quality. Although RCTs are usually cited as the highest level of evidence for judging the efficacy of therapeutic interventions, randomization should not be seen as a reliable proxy for overall quality.[18] Well-conducted nonrandomized studies may be more valid than poorly conducted RCTs. In this review we found 2 RCTs[17;48] with a lower score on the criteria for internal validity than we accepted from other designs. Another unexpected finding was that 4 RCTs[48-51] and 4 CCTs[28;52-54] did not present between-groups comparisons. We recommend the presentation of between-group comparisons to be an additional criterion in the future rating of methodologic quality.

The various types of NMD and PT interventions in combination with the use of a variety of outcome measures required that decisions were made on classifications and cutoff points. We decided that more than half of the outcome variables in a given study had to show a significant effect in order to

provide evidence. We presumed that all outcome variables were of similar importance, but this is arbitrary. It may have resulted in loss of evidence when effects of important variables were overruled by other variables without an effect. However, when we checked the actual variables, they were generally in agreement with our assumptions.

Finally, in our method, a study that provided evidence of effectiveness, could be neutralized by studies without substantial evidence. One can argue that this is too strict, considering the fact that a study could only provide evidence if more than half of the variables showed a significant effect. On the other hand, we were not strict in including all RCTs and CCTs regardless their methodologic quality and other designs of sufficient methodologic quality.

Exercise intensity

Most studies evaluated muscle strengthening exercises, aerobic exercises, or a combination of these. The intensity of strengtening exercises can be manipulated by varying the resistance or weight, the number of repetitions, the length of the rest interval, or the number of sets of exercises completed. The American College of Sports Medicine (ACSM) formulated minimal requirements to evaluate the quality of training programs for effective muscle strength training in healthy adults[80]. The ACSM recommends a progressive individualized program, for all major muscle groups with at least 1 set of 8 to 12 repetitions and a frequency of 2 to 3 days a week. Obviously, these requirements for healthy individuals cannot simply be applied to persons with NMD, because of lack of evidence regarding the effect of physical exertion on the diseased neuromuscular system. Overexerting muscles might accelerate disease progression[12]. Still, the intervention should be of enough intensity in order to provide a training stimulus.

Nearly all studies on muscle strengthening exercises included in this review met the requirements of an individualized and progressive program. However, there was considerable variation in the muscles exercised, type of exercises given, and type of resistance, number of repetitions, and number of sets. Most investigators used a moderate level of intensity in order to prevent adverse effects. Indeed, a very important finding was the absence of adverse effects. If, however, the intensity is too low, one cannot expect an effect of training other than physiologic adaptations due to activation of muscles that might have been inactive before.

For aerobic exercises, the ACSM recommends the use of large muscle groups in a rhythmic, aerobic and continuous manner[18;81]. For most people,

intensities within the range of 70% to 85% of maximum heart rate, or 60 to 80% of oxygen uptake reserve or heart rate reserve are sufficient to achieve improvements in cardiorespiratory fitness, when combined with an optimal frequency of 3 to 5 days a week [81]. This review has shown that these intensities can be recommended for patients with NMD without adverse effects. Most of the included studies used cycling or treadmill exercise with a frequency of at least 3 times a week and exercise intensity around 70% of heart rate reserve or estimated maximum heart rate.

For both muscle strengthening and aerobic exercises the entire program should last at least 10 weeks and regular supervision optimizes the effect of training and improves safety and compliance[12]. Yet, in only 30% of all studies on muscle strengthening and aerobic exercises, the duration of the interventions was less than 10 weeks and in approximately 50% of the studies, the training was under regular supervision.

Uniformity

In order to facilitate meaningful comparisons among studies and statistical power by effective pooling of study results, more uniformity is needed in type of interventions, intensity of exercise therapy and type of outcome measures. To achieve more uniform terminology, we recommend the development of an international classification for PT modalities. In the Netherlands, such a classification has been developed for allied health care professionals[82]. In this study, the ICF[19] was used to make a distinction between outcome variables at the level of body functions and at the level of activities and participation. We recommend to develop ICF core sets specifically for NMD, like ICF core sets developed for other chronic diseases[83-85]. This would provide professionals and researchers with a framework for the selection of assessment and outcome measures on body functions, activities and participation and on environmental factors for research as well as for clinical purposes.

Conclusions

Our best evidence synthesis resulted in level II evidence (likely to be effective) for strengthening exercises in combination with aerobic exercises for patients with muscle disorders. Level III evidence (indications of effectiveness) was found for aerobic exercises in patients with muscle disorders and for the combination of muscle strengthening and aerobic exercises in a heterogeneous group of muscle disorders. Finally, there is level

III evidence for breathing exercises for patients with MG and for patients with MMD. There was insufficient evidence for strengthening exercises due to insignificant or inconsistent effects. Most studies reported the absence of adverse effects.

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Part II: Scientific perspective

Chapter 6
Speech pathology interventions in patients with neuromuscular diseases: a systematic review

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Abstract

Purpose: A systematic review was conducted to summarize and evaluate the literature on the effectiveness of speech pathology interventions in adults with neuromuscular diseases.

Method: Databases searched included the Cochrane Database of Systematic Reviews, the Cochrane Central Register of Controlled Trials, Medline, CINAHL, Embase, PsycINFO and PubMed. A total of 1,772 articles were independently screened on title and abstract by 2 reviewers.

Results: No randomized controlled trials or clinical controlled trials were found. Four other designs were included. Only one study on oculopharyngeal muscle dystrophy (OPMD) appeared to have sufficient methodological quality. There is evidence indicating that correction of head position in patients with OPMD improves swallowing efficiency (level III evidence).

Conclusion: Despite 1,772 studies, there is only evidence of level III regarding the effectiveness of speech pathology interventions in patients with OPMD. Recommendations for future research are given.

Introduction

Neuromuscular diseases (NMD) represent a subfield of neurology, including heterogeneous groups of patients with approximately 600 diseases of the muscle, neuromuscular transmission, peripheral nerve and nerve root or motor neuron. NMD can lead to dysarthria and dysphagia, which are both diagnosed and treated by speech pathologists (SP).

Both dysarthria and dysphagia are not only relatively prevalent, but also clinically relevant symptoms in NMD. The impact of dysarthria on quality of life is obvious, since communication is generally considered to be a critical determinant of quality of life. Dysphagia can lead to aspiration pneumonia and therefore to serious health problems. Dysphagia and concomitant pneumonia may even be life-threatening in otherwise weakened NMD patients.

In a sample of 102 NMD patients, we found that 45% of the patients had an indication for SP advice, regarding dysarthria or dysphagia[1]. However, this judgement was based on the expertise of the participating SP. The aim of this article is to assess whether there is evidence for the efficacy of treatments administered by SP in patients with NMD.

Methods

Search strategy

We searched the Cochrane Database of Systematic Reviews and the Cochrane Central Register of Controlled Trials (The Cochrane Library November 2007, Issue 3), Medline (1966 through November 2007), CINAHL (1982 through November 2007), Embase (1980 through November 2007), PsycINFO (1806 through November 2007) and PubMed (1950 through November 2007).

The search strategy was built on different types of NMD and different types of SP interventions. For all search strategies Medical Subject Headings (MeSH) or indexed terms were used as well as free text words. The full search strategy is shown in Table 1. Reference lists of reviews and selected articles were scanned for further potentially relevant articles.

Selection criteria

Inclusion was restricted to peer-reviewed articles published in English, German, French or Dutch. Randomized controlled trials (RCTs), clinical controlled trials (CCTs) and other designs (ODs) were included. Single case studies were excluded.

Table 1. Free text words and MeSH terms used in search strategy to find evidence for the effectiveness of speech pathology in NMD

Free text words
Speech pathology
Speech pathology, speech therapy, speech and language therapy, speech disorder, communication disorder, articulation disorder, swallow disorder, deglutition disorder, dysphagia, dysarthria, dietary modification, swallow, intelligibility, alternative communication
NMD
Neuromuscular disease, neuromuscular disorder, muscle disease, muscle disorder, neuromuscular junction disease, neuromuscular junction disorder, motor neuron disease, motor neuron disorder, motoneuron disease, motoneuron disorder, neuropathy, polyneuropathy, peripheral nervous system disease, peripheral nervous system disorder, neuralgia, neuritis, myopathy, dystrophy, myotony, myositis
MeSH terms
Speech pathology
Dysarthria, speech disorder(s), speech therapy, speech pathology, communication disorders, communication aids, speech intelligibility, facilitated communication, communication aid, alternative and augmentative communication, communication aids for the disabled, dysphagia, deglutition disorders
NMD
Neuromuscular disease(s), neuromuscular disorders

Participants included adults (>18 years) with various types of NMD (disorders of the muscle, neuromuscular transmission, peripheral nerve and nerve root or motor neuron). SP interventions included information and advice or teaching compensatory strategies. Information and advice incorporated (1) dietary modification, (2) augmentative and alternative communication, or (3) instruction of the patient and relatives. Compensatory strategies included teaching swallowing maneuvers and/or strategies to improve intelligibility. Brain computer communication devices such as thought translation devices were excluded, as this is not considered a regular SP intervention. For the same reason, we also excluded medical interventions related to dysphagia, such as myotomy of the cricopharyngeal muscle.

The outcome measures included swallowing at the level of body functions and intelligibility, communication and eating and drinking at the level of activities and participation, according to the definitions of International Classification of Functioning, Disability and Health (ICF)[2].

Procedure for inclusion

Two reviewers (SK and EHCC) independently carried out a preliminary selection based on title and abstract of articles. Full texts of articles that seemed to fit the inclusion criteria were retrieved for further evaluation. A final selection was made independently by the 2 reviewers on the basis of full texts. When different conclusions were drawn, consensus was reached through discussion.

Methodological quality

Methodological quality of studies was independently rated using criteria recommended by Van Tulder et al.[3]. The 2 reviewers independently assessed the methodological quality. Disagreements were resolved by discussion between the reviewers.

For RCTs and CCTs, the *quality of reporting* was scored on 6 criteria (eligibility criteria, comparability of groups, explicit description of interventions, adverse effects, short-term and long-term follow-up measurements). *Methodological quality* was scored on 9 criteria (randomization, concealed allocation, avoidance of co-interventions, compliance, blinding of outcome assessor, relevance of outcome measures, acceptable dropout rate, comparable timing of outcome assessment and intention-to-treat analysis). *Statistical quality* was scored using 2 criteria (description of sample size and presentation of point estimates/measures of variability). One criterion regarding statistical quality (between-group statistical comparison) of the PEDro scale was added[4].

Methodological quality of articles was considered sufficient when 3 out of 6 reporting criteria, 6 out of 9 methodological criteria, and 2 out of 3 statistical criteria were scored positively[3]. For ODs, the criteria of comparability of groups, randomization, allocation, and between-group comparison were not applicable. For these designs, 2 out of 5 reporting criteria, 4 out of 7 methodological criteria, and 1 of 2 statistical criteria had to be scored positively to be of sufficient quality[5].

Best evidence synthesis

A best-evidence synthesis was performed according to the classification of the Dutch Institute for Health Care Quality Improvement[6]. Three levels of evidence and conclusions were formulated. Level I evidence refers to at least 2 RCTs of sufficient quality. Conclusions are formulated as 'it has been shown that....'. Level II evidence refers to 1 good quality RCT or at least 2 independent controlled studies (RCTs or CCTs) of less methodological

quality. Conclusions are formulated as 'it is likely that....'. Level III evidence refers to an RCT or CCT of low methodological quality or at least 1 OD of sufficient methodological quality. Conclusions are formulated as 'there are indications that....'. When inconsistent findings were found in studies of similar design and methodological quality, conclusions are formulated as 'there is insufficient evidence that....'.

Results

Selection

The search strategy identified 1,772 articles, duplicates excluded (Figure 1). After screening on title and abstract, 1,723 articles were excluded because they did not meet predefined criteria for study design, type of participants, type of intervention, type of outcome measures, or English, French, German or Dutch language. In 42 articles there was discussion between the 2 reviewers, leading eventually to retrieval of 17 full-text articles that seemed to fit the inclusion criteria. In addition, 7 review articles were retrieved and screened for additional articles. Another 15 articles from reference lists were retrieved in full text. Following evaluation of the full-text articles, 4 fulfilled all inclusion criteria[7-10]. These studies were subjected to assessment of methodological quality.

Methodological assessment

The 4 studies included were all ODs. The methodological quality of 3 studies was insufficient (8-10) and the methodological quality of 1 study was found sufficient[7] (Table 2).

De Swart et al.[7] found a negative influence of ptosis on swallowing function in patients with oculopharyngeal muscle dystrophy. The instruction 'head position slightly flexed', i.e. head not adapted to the ptosis, significantly increased swallowing efficiency. This was objectively evaluated with videofluoroscopy (with 20 ml of thin and thick liquid) and maximum swallowing volume.

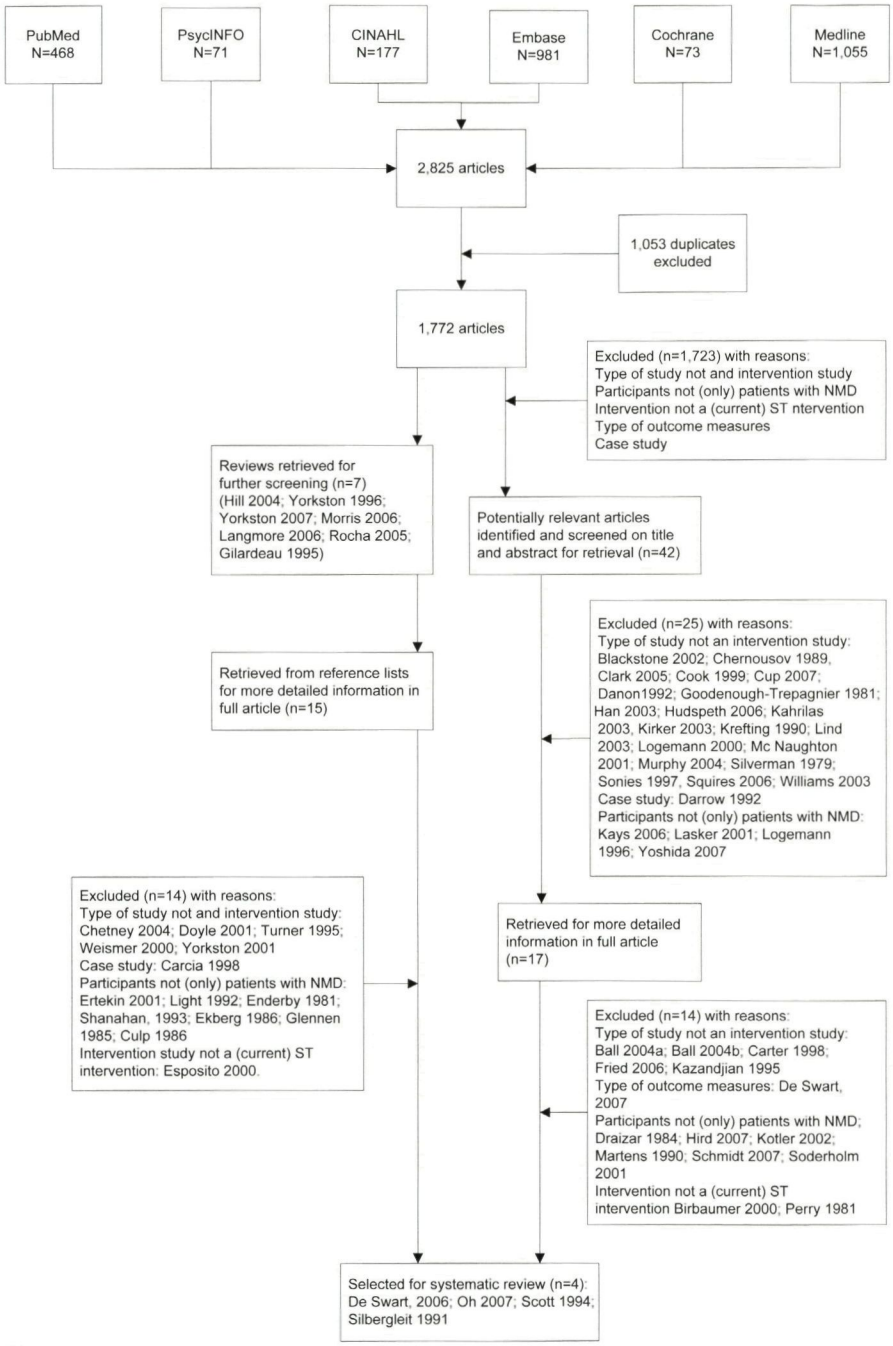

Figure 1. Process for identification of included studies.

Table 2: Methodological quality of included studies showing 3 studies of insufficient methodological quality (8-10) and 1 study of sufficient methodological quality[7]

Criteria	Silbergleit et al.[10]	Oh et al.[8]	Scott & Austin[9]	De Swart et al.[7]
Reporting criteria				
Eligibility criteria specified?	Yes	Yes	Yes	Yes
Intervention explicitly described?	Yes	Yes	No	Yes
Adverse effects described?	No	Yes	Yes	No
Short-term follow-up measurement performed?	No	No	Unknown	Yes
Long-term measurement performed?	Yes	Yes	Yes	No
Total descriptive criteria (at least 2/5)	3/5	4/5	3/5	3/5
Methodological criteria				
Co-interventions avoided?	Unknown	No	No	Yes
Compliance acceptable?	Yes	Unknown	Yes	Yes
Outcome assessor not involved in the treatment?	Unknown	Unknown	No	N.A.
Outcome measures relevant?	Yes	Unknown	No	Yes
Withdrawal/drop-out rate described and acceptable?	Yes	No	Yes	Yes
Timing of the outcome assessment in all patients comparable?	No	No	Unknown	Yes
Analysis including an intention-to-treat analysis?	No	No	Unknown	No
Total internal validity (at least 4/7)	3/7	0/7	2/7	5/7
Statistical criteria				
Was the sample size of the group described?	Yes	Yes	Yes	Yes
Were point estimates and measures of variablility presented for the primary outcome measures?	Yes	No	No	Yes
Total statistical criteria (at least 1/2)	2/2	1/2	1/2	2/2

Best-evidence synthesis

The study by de Swart et al.[7] is an OD of sufficient methodological quality. Therefore the evidence-based conclusion was that there are indications (level III evidence) that when head position is slightly flexed and not adapted to the ptosis, swallowing efficiency improves in patients with oculopharyngeal muscle dystrophy.

Discussion

The main conclusion of this study is that the published evidence for effectiveness of SP in adults with NMD is very limited, as our search strategy did not identify RCTs or CCTs on this topic. There was only 1 OD (uncontrolled pre-post study) that was of sufficient methodological quality.

Regarding dysphagia, the only available evidence (level III) found was based on an uncontrolled study[7]. As we know from normal swallowing, extension of the neck leads to ineffective swallowing[11-13]. Therefore, basic instructions about the influence of posture of head and neck on swallowing seem valuable in NMD in general – particularly when the disease leads to changes in posture or weakness in the head-neck-shoulder region or when the disease leads to weakness in muscles involved in speech or swallowing.

Regarding dysarthria, despite the extensive amount of articles, no evidence was found for the effectiveness of SP in NMD. We only found 2 articles in which one-time manipulations of speaking rate led to a slower speaking rate, but this technique did not have a positive effect on intelligibility.[14;15].

All 4 studies included a homogeneous patient population with regard to the medical diagnosis. This may limit generalization of the results to other groups of patients with NMD. However, many NMD patients suffer from weakness in speech and swallowing muscles. Therefore, we hypothesize that the SP interventions are applicable to different groups of NMD patients with comparable impairments and disabilities. The comparison between studies is also difficult because of the different outcome measures used. Finally, considering the small number of patients, there might be a type II error in the included studies. This problem can be solved by including different groups of NMD patients with comparable impairments in one study.

Exclusion of studies

Our extensive search resulted in a large number of articles, most of which were excluded. Many studies did not evaluate the effectiveness of an SP

interventions but were descriptive studies, for instance on dysarthria and dysphagia in patients with NMD. The use of MeSH and free text words for different types of NMD resulted in many studies concerning other neurological conditions, such as Parkinson's disease or stroke, which did not fulfill our inclusion criteria.

Recommendations for future research

Despite their potential limiting effect on quality of life and general health condition, there are hardly any studies focusing on the effects of dysarthria and dysphagia. We would like to propose that both dysarthria and dysphagia are of utmost importance in NMD patients and treatment strategies with respect to these impairments should be the topic of well-designed RCTs. Preferably, these trials should be performed in homogenous patient samples, with comparable levels of impairments. Conclusions of those trials might be used in other groups of NMD patients, with similar impairments at the bulbar level. However, these remarks should not discourage researchers to perform less ambitious research, for example observational cohort studies or controlled trials. Results of such trials also add to the required level of evidence of SP.

As for the interventions, it is important that all patients are given standardized interventions and that SP interventions are being properly described. It should be noted that the feasibility of interventions depends on the type of intervention. Swallowing techniques are internationally uniform and well described in the literature[16]. However, there is less uniformity regarding techniques to improve intelligibility, although interventions like decreasing speaking rate or producing a louder voice can be objectively described in syllables per second or by measuring sound levels. Interventions regarding augmentative and alternative communication and instruction of the patient and relatives are tailor-made to such an extent that they cannot be standardized.

With regard to the type of outcome measures, we recommend using videofluoroscopy or flexible endoscopic evaluation of swallowing to objectively evaluate swallowing. Also, other quantitative measurements like the timed test[17] and the dysphagia limit[18] can be easily used as objective outcome measures. Secondary outcome measures like body weight, percentage of oral intake and quality of life can be valuable. For evaluating speech at the level of activities, we recommend using a speech intelligibility test[19-21]. At the level of participation, a scale of communicative participation is being developed[22]. Acoustic measurements are the most objective

measurements to evaluate speech at the level of body functions. It should be kept in mind, however, that acoustic measurements have limited predictive value with regard to communication effectiveness[23-25]. Therefore we recommend using the acoustic measurements only in combination with measurements at the level of activities or participation.

A consensus meeting with experts on speech pathology, neurology and methodology is recommended to further discuss and agree on the outlines of future trials. Previous meetings along these lines and covering various neuromuscular topics, such as myotonic dystrophy, have been very succesful[26] .

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Part III: Client perspective

Chapter 7
Living with myotonic dystrophy; what can be learned from couples? A qualitative study

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Abstract

To increase understanding of how myotonic dystrophy type 1 (MD1) affects the lives of couples and how they manage individually and together, a qualitative study was carried out. Fifteen in-depth interviews with five couples took place in their homes. People with MD1 associate this neuromuscular disease with decreasing abilities, describing physical, cognitive and psychosocial barriers to everyday activities and social participation. Partners highlighted the increasing care giving burden, giving directions and using reminders to compensate for the lack of initiative and avoidant behaviour due to MD1. Couples portrayed the dilemmas and frustrations of renegotiating roles and responsibilities; stressing the importance of achieving a balance between individual and shared activities. All participants experienced a lack of understanding from relatives, friends, and society, including health care, leading to withdrawal and isolation. Couple management, supporting the self-management skills of both partners is proposed as a way to better match health services and needs.

Introduction

"Every time I ask her to come shopping with me, she says you better go without me".

This quote is typical for a partner of someone with myotonic dystrophy type 1 (MD1). It may raise questions like why this person with MD1 chooses to stay at home? What does it mean for her and her partner? Is it considered a problem or not and if so by whom? These questions address the impact of MD1 on daily life of couples and are related to their health and well-being. How often are such issues addressed by health care professionals?

MD1, also known as Steinert disease, is the most frequent of the adult neuromuscular diseases with a prevalence estimated at 1/20,000 inhabitants[1]. MD1 is a hereditary chronic systemic disease with wide variability in clinical expression, both within and between families. MD1 is characterized by the involvement of many organs, including impairments in the muscular, respiratory, cardiac, central nervous, endocrine and ocular functions and structures[2-4]. Typical symptoms of the disease include myotonia and progressive loss of muscle strength, usually distal to proximal, and weakness of facial and anterior neck muscles. Other symptoms include cataracts, frontal baldness, dysarthria, fatigue and daytime somnolence. Also cognitive decline including lack of initiative and specific personality traits such as an avoidant personality are related to MD1[5-8]. Compared to other neuromuscular diseases, persons with MD1 have among the gravest functional disabilities and the greatest dependence on others for activities of daily living[9]. They also have the lowest social participation, most psychosocial problems and poorest psychosocial well-being[10]. This not only has huge impact on persons having the disease[8;10;11], as well as on their partners [12;13] and surely on their lives together as a couple. As far as we know, this is the first study to explore the impact of MD1 on the daily lives of couples.

Despite the huge impact of MD1 on daily life, individuals with MD1 tend to play down or avoid expressing their problems when attending physicians or other health care professionals and adherence to rehabilitation services is generally poor [14]. Reasons for this are not well understood. Difficulties managing their own health has been attributed to personality features such as diminished persistence and increased avoidance[5;15;16]. It may also be that persons with MD1 perceive their limitations as normal, since the slowly progressive nature of the deterioration gives them time to adapt and live more by what they can do than by what they cannot do [10] or they might have received the message that the disease is progressive and that there is no

cure[8]. Finally, they may believe that professionals cannot help and have little knowledge and understanding of the disease[8]. Indeed, although MD1 is one of the most prevalent neuromuscular diseases, the disease is relatively unknown among medical and allied health professionals and scientific evidence of the effectiveness of allied health care is limited as shown in the various systematic reviews[17-19]. Very little is known about how couples cope in daily life with this complex, progressive disease. It is important to understand their perspective of living with this hereditary, systemic disease in order to better match health care to their problems, concerns and needs. Persons with MD1 and their partners both live with the disease and become experts through their experience. They are more knowledgeable than professionals regarding the consequences of the disease for their lives and how treatment effects their daily lives[20;21].

The purpose of this study is to increase our understanding of what MD1 means for persons having the disease, for their partners and for both as a couple, and how they manage in daily life individually and together. The research questions are:

- How do persons with MD1 and their partners reflect on living with this hereditary chronic progressive disease?
- How do persons with MD1 and their partners experience their lives together as a couple?

Methodology

Design

As we aim to study personal experiences, make sense of these and interpret these in terms of the meanings the persons themselves bring to them, a qualitative method is most appropriate[22]. Couples were visited in their homes. In 2009, 15 in-depth interviews were carried out with persons with MD1 and their partners separately and simultaneously at two different moments in time to obtain insights in the meaning of MD1 on the lives of couples.

This study followed a hermeneutic approach according to the method of '*verstehen*', which is based on three principles[22]:

1) Human experience has a historical nature and is context bound. This was addressed by asking the participants about present and past experiences and the context of these and by conducting the interviews in the homes of the couples, their naturalistic setting.
2) Understanding requires imaginative participation in the lives of those being studied to not impose foreign meanings on their thoughts and

actions. This was addressed by listening to the couples and observing how they live and manage in their home situation.

3) The researchers' professional perspectives and experiences are used when interpreting the findings to gain better insight and understanding of the meaning of living with MD1.

Data collection and procedure

The researcher (EC) approached all couples by telephone. All agreed to receive written information about the study. When contacted a week later, all couples agreed to participate in the study. The first couple served as a pilot couple to test the interview guide (Box 1). As lack of understanding was a major issue raised spontaneously by this couple, this topic was added to the interview guide.

Data collection was iterative and consisted of four phases for each couple:

1) Interview with the person having the MD1;
2) Interview with the partner;
3) Verbatim transcription of interviews and selection of meaningful expressions or topics by the researcher (EC);
4) Interview with couple about three weeks later. The goal was to elaborate on previous topics and to get more in depth information on how MD1 affects the lives of couples and how they manage together.

Preceding the interviews, informed consent forms were signed and all participants gave permission to record the interviews. Small talk and an explanation of the purpose and format of the interview enabled the interviewer and interviewees to become acquainted with each other. The interview guide comprised one main question and follow-up topics. Each interview proceeded in an informal conversational style, lasting between 60 and 90 minutes. Following the interviews, the researcher wrote memos on the impressions and reflections of the interview and field notes about the home situation.

Ethical considerations

At all visits the researcher emphasized that participation was entirely voluntary and that the participants could withdraw at any time without consequences for their treatment. Permission to audiotape the interview was confirmed at every visit as was reassurance of confidentiality. Following the interviews, the researcher gave participants an opportunity to reflect on the experiences and checked the need for formal after care from a social worker.

None of the participants indicated they needed this. All data and quotes used were anonymized. Approval from the local medical ethics committee (CMO number 2008/261 NL26868.091.09) was obtained.

Box 1. Interview guide
Main question:
"How has myotonic dystrophy influenced your daily life and how do you manage?"

Topics:
Current daily activities, roles and relations and their meaning
Activities, roles and relations and their meaning in the past
Changes in activities roles and relations
Management strategies to deal with changes

Topic added following the pilot interview:
Understanding from others

Participants
Ten persons, five with MD1 (adult or classical type) and their partners, were included. Purposive sampling [23] was used, identifying couples with MD1 from the Dutch neuromuscular database, CRAMP (Computer Registry of All Myopathies and Polyneuropathies) which contains about 300 persons with MD1. All participants with MD1 had been admitted to the medical centre in the past for the purpose of multidisciplinary (medical and allied health) assessment and advice. Criteria for inclusion were: 1) the person with MD1 was restricted in activities of daily living and social participation; and 2) the couples had been living together for at least ten years. Difficulties in communication such as dysarthria, which had been an exclusion criterion in other qualitative studies with persons having MD1 only, was not an exclusion criterion in the current study.

Three men and two women with MD1 and their partners, all Caucasian, representing the majority of couples with MD1 in the Netherlands, were willing to participate. They lived in different villages in the south of the Netherlands. The mean age of the persons with MD1 was 58 years (range 53 – 65). There was variation in socio-economic background and work status. One of the persons with MD1 still worked part-time in a sheltered work facility, two were retired and two were housewives. One partner worked

fulltime, one part-time, one worked on occasions and one partner had taken early retirement. Two couples had one or more children with MD.

Data analysis

Analysis started following initial interviews with each couple. In depth analysis took place after completing all 15 interviews. Analysis involved a constant comparison method[24] including the following steps:

1) Familiarization with the interviews and understanding of the experiences and meaning of living with MD1 by listening and multiple readings of the transcripts, the production of summaries and reading the reflexive memos and field notes;

2) Assignment of codes to meaningful text units (statements) related to the research questions. In-vivo codes were assigned to stay close to the original voices of the interviewees;

3) Categorization of meaning units from the combination of all codes, reorganization and interpretation, resulting in the formulation of themes and subthemes;

4) Interpretation of relations between themes and subthemes with use of other sources including literature, field notes and own experiences leading to new insights and better understanding of the meaning of MD1 for couples.

The researcher (EC) performed all steps and two co-authors (TS, AK) served as peer-reviewers during the process of coding and the formulation of themes and subthemes. The codes, themes and subthemes were discussed with the whole research group at three meetings. MaxQDA 2007 computer software was used to assist in organizing the data (www.MAXQDA.com). The analysis was carried out in the Dutch language to minimize bias and preserve meaning. Only the quotations used in this article were translated into English.

Reflection on the role of researcher and research group

The research group included researchers with different medical, philosophical, allied health care and methodology background. Three members were experienced researchers and lecturers in the field of qualitative research (AK, TS, GJW). The researcher (EC) also worked as an occupational therapist (OT) in a multidisciplinary team of clinicians and researchers. Previously, she had seen three of the participants with MD1 for a single assessment and advice in the neuromuscular centre. In the restricted time allocated in a clinical setting, she had felt it was impossible to understand what it is like for couples living with MD1. The fact that the

researcher was known to three people with MD1 did not seem to influence the participants in sharing their personal experiences.

Trustworthiness

To further enhance the trustworthiness or rigor of the study[25], several triangulation methods were used[26], including the use of different perspectives (persons with MD1, their partners and as a couple) generating richer understanding than single perspectives[27]; and investigator triangulation within the research group. Credibility[25] of the study was further ensured by member checking and the use of participants' quotes and in-vivo (sub) themes. All couples had received summaries of the interviews including meaningful quotes and had confirmed content.

Box 2. The four themes resulting from the analysis

Impact for persons with MD1 (35% of codes):

1) Decreasing abilities

- Many barriers; "no power, no pep, no guts"
- Avoiding barriers; "I don't do that much"

Impact for partners (15% of codes):

2) Increasing burden

- "If I did not do it, nothing happened"
- "I am in charge; I give directions"

Impact for couples (25% of codes):

3) Finding a mode together

- Doing or leaving; renegotiation of tasks
- Individual and shared activities; give each other freedom

Impact for persons with MD1, for partners and for couples (20% of codes):

4) Lack of understanding

- Even the family does not understand
- We manage ourselves
- Health care: many islands focused on part of the disease

Results

In total 1070 codes were assigned to meaningful text units in the transcribed interviews. Of these, most codes (433) came out of the interviews with the partners and 339 codes were from the interviews with persons with MD1.

The couple interviews yielded another 298 codes. Four themes emerged representing the impact for persons with MD1 (35% of the codes), for their partners (15% of the codes), for couples (25% of the codes), and the last theme represented an overall impact (20% of all codes) (Box 2). The subthemes reflect the impact of living with MD1 and the main self-management strategy used. To capture the essence of a (sub) theme, in vivo codes were used.

Theme 1: Decreasing abilities

Participants with MD1 all experienced progressively more limitations in an increasing number of daily activities and roles which were important for them. These activities were enjoyable, made them independent, gave a sense of belonging or made them feel valued by others. For some participants the decrease in abilities felt like "I cannot do anything anymore".

> "I have always helped my brothers a lot. Those were good times. I could do it all. But that's history. I cannot do anything anymore. Now they no longer need me. They have sort of forgotten me."

Many barriers; no power, no pep, no guts

Participants experienced obstacles to everyday activities and social participation. Some referred to barriers; others said it was too much trouble, too much hassle, too difficult, or too complicated. When someone said that he or she lacked pep or guts, it could mean that he or she lacked energy or vitality (physical barrier), experienced difficulty initiating or planning tasks (cognitive barrier), or lacked courage (psychosocial barrier) or a combination thereof.

> "For a lot of things I just don't have the guts. Then it takes ages before I get dressed, simply because I don't feel like it."

Participants reported physical limitations such as reduced strength in hands and legs, decreased balance, reduced fitness and fatigue. These limitations led to safety risks, fear of falling, inability or a disproportionate amount of time or effort was needed to perform tasks.

> "I used to make the table. But I cannot do this anymore. Because, when I take the plates and I fall, then we have no plates anymore ..."

> "My condition is nearly nothing. I cannot walk anymore, at least, not what it should be. When I walk, I'm dead tired. I walk as little as possible."

Several participants mentioned problems with their bowels or continence which restricted participation in daily activities.

> "I used to sit on the toilet the whole morning. Now I notice with my bladder sometimes..... I cannot do much, otherwise I may lose control"

Few participants described lack of initiative or mentioned that thinking what to do or how to do it was difficult for them. Instead, many regarded themselves as 'easy' and some wondered whether they might be too easy going.

> "Deciding what we are going to eat is terribly difficult for me".

> "Persons with my muscle disease don't have much initiative. If my wife suggests shall we go on holiday? Then I say okay, you say where, because I don't care. She does not like that. She says you can think of what you like. Then I say I enjoy everything, I am very easy."

In social situations, persons with MD1 found it hard to take initiative and usually waited for invitations from others. They described barriers associated with feelings of shame or insecurity. The reduced intelligibility or clumsiness made them feel anxious and led to fear of being looked at or being made a fool of.

> "On a birthday, I'll quietly sit in a corner. I dare not say anything. There are also strange people that might not understand me or make me feel foolish."

Avoiding barriers; I don't do that much

Persons with MD1 mentioned that they increasingly give up, postpone or avoid activities and roles, like stopping work or sports and going out less. Even the daily shopping is often left to the partner. The result is that their world becomes smaller and social contacts decline. Although this is perceived as a difficult and painful process, the participants felt that there is no use in complaining and tried to stay positive.

> "I get very annoyed when I'm in a shop and I cannot open a bag to put tomatoes in it or something. Then I have to ask to keep the bag open. It may

be false modesty or shame, but I find that terrible. That's why I have quitted doing the shopping."

"Because you are at home most of the time, your world becomes smaller, which is very annoying for me..... But I am not going to sit and be sad. I still do things, but not that much."

Participants also adjusted their expectations and demands, making life more manageable. Sedentary or spectator activities were mentioned like enjoying the presence of grandchildren or sitting in the garden or at the camping site. They described adapting activities or doing less complicated activities in order to participate with less effort.

"I'm short of breath. When I sing, I occasionally cheat, I call it playback. We have performances, which is nice and it is also good for social contacts. "

Participants found alternatives like playing bridge instead of tennis or joining a book club instead of the quilt club. Being a member of a club or a group of friends with regular appointments appeared very helpful to stay in touch with others and have fun. Participants mentioned that they perceived no barriers when doing sedentary activities such as reading or watching television and experienced much pleasure doing this.

"Television is important to me. Especially since I've always been a sports fan, I like to watch sports. Soon Wimbledon and Tour de France are on television. Then I watch television in the afternoon. For me it's an important thing to relax."

Theme 2: Increasing burden

Partners expressed how they gradually had taken over more activities or even the entire organization of their lives as a couple. Because it happens gradually, it is experienced as an ordinary natural process, but also as a burden.

"You feel that you are on your own and have to do it all. The idea that it will never change, can be an enormous burden". [partner]

All partners mentioned how the disease effected their employment. Most partners had been able to adapt their working hours to their home circumstances, combining home and work tasks to support their partner.

If I did not do it, nothing happened

Partners experienced that they had to do it all: the household, the administration as well as initiating social events and keep in touch with friends and family. They noticed the difficulties their partners with MD1 had in initiating action. The inactivity and deferral of duties often was a frustrating experience for partners.

> "What can be postponed was postponed. It completely irritated me. Then I thought come on, do something!" [partner]

I am in charge; I give directions

Partners found themselves completely in charge of what needs to be done, whereas in the past they had shared responsibilities. Several partners used reminders such as writing down duties or making a telephone call from work to prompt actions. Partners noted the willingness of the person with MD1 to act, but the need for an extra stimulus to actually get going.

> "I am often the one who plans everything. He usually agrees. Sometimes he is too tired for things. Then I have to say well come on. I write down the things he can do. I say you're at home all day, so you can also do something, stay busy, we must do it together." [partner]

Some partners deliberately gave directions based on their knowledge and understanding of the cognitive consequences of MD1. However, not all partners were aware of these. One partner felt that her partner's brain did not work well, as if there was some kind of dementia. She felt that she constantly had to say something, otherwise nothing would be done.

Theme 3: Finding a mode together

Couples experienced living with MD1 as a process with dilemmas and frustrations, but compared it to the give and take required in every marriage. They felt ambivalence whether to do tasks themselves or encourage their partner to do it. Giving each other freedom and respecting differences in pace and interests were seen as important ways of managing together.

> "I think it is important that you do it together. The one with the disease should not give up too easily, but should try to do what is possible. You have to find a good mode together." [partner]

MD1 leads to changes in roles and the marital relationship. Several partners indicated that they can imagine that marriages fail. A partner told how she had been jealous on other couples who happily did things together, while her husband always fell asleep. When she was 40, she felt being married to a man of 80. Couples also had to deal with changes in sexuality and intimacy in the relationship, which both experienced as difficult.

> "You become a carer". [partner]
> "We pretty much live like brother and sister."

Doing or leaving; renegotiation of tasks

Participants with MD1 said they want to do things themselves for as long as possible. They experienced that their partner sometimes gets impatient when performance of tasks takes much time. On the other hand they welcomed partner assistance when efforts became disproportionate for them or associated with safety risks. Partners faced dilemmas when to encourage their partner to do activities themselves and when to take over. Couples indicated the importance for the person with MD1 remaining active even though doing so, demanded more time, effort or was clumsy.

> "If it is difficult to rise from your chair because you have pain in your legs and your arms and you want to put on the potatoes. Then it is much easier to ask the other to do it..... For things I know that she cannot do it, I have no problems to take over. I get annoyed when I think if you try a little harder, you can do it yourself". [partner]

The balance between encouraging and taking over seemed to be variable and context-specific. A partner mentioned how she enjoyed watching her husband playing with their granddaughter. She knew this cost him energy, so she took over other activities for him. Previous roles and personal interests influenced this balance. In some couples there was consciously renegotiating of tasks, but there were also couples whereby nearly all tasks were gradually taken over by the partner.

Individual and shared activities; give each other freedom

Couples experienced an increasing difference between each other's capacities, pace and interests. They thought about what they could do together and what they could not. If they choose 'together', they adjusted the pace and enjoyed being engaged in shared activities.

> "If we go shopping together, we do this leisurely at a snail's pace. Then it is not that I run to the supermarket after work, toss something in the car and fly home. No, we take the time and enjoy shopping together. We have chosen to do it this way." [partner]

All couples commented that going out was important for them. However, spontaneous outings or holidays had led to disappointment due to inaccessible public buildings. This meant that outings increasingly needed planning and preparation, usually done by the partners. Limited stamina, fatigue and sleepiness of persons with MD1 also influenced going out together because of the rest needed during the day.

> "If we go away together, we are often confronted with a lot of problems. If you go to a museum, you might have to take six flights of stairs before reaching the entrance, then the game is over for us. ... It means that you cannot go anywhere spontaneously." [partner]

Couples found solutions to engage in shared activities, like cycling with electric bikes in the surroundings, visiting accessible restaurants or taking a wheelchair to go out. Several spoke about enjoying just sitting together in their garden. Couples emphasised the importance of giving each other freedom. Persons with MD1 were usually quite happy for their partner to do things for themselves. Partners learned that doing things for themselves is a way to re-energise and was experienced as taking good care of one self.

> "I give my wife a lot of freedom. If she wants to go away with friends, that's fine with me. You go and have fun, I don't feel the need to go."
> "Freedom is important, because at times it is quite heavy. A source of energy, whether it is sports or something else, gives you new energy, as long as you have something for yourself." [partner]

Theme 4: Lack of understanding

Persons with MD1 reported feeling misunderstood and judged as silly or sulky, due to their facial weakness. Couples described a lack of understanding within their family, among relatives and friends and in the health care system and society at large. They speculated that was this was due to invisible problems like fatigue, but felt hesitant to ask for understanding, tending to withdraw from situations and hide their problems.

"Previously, I was the centre of attention. That is no longer the case and I have to get used to that. Now I am being looked at as if I am 'not quite in line'. I notice this and it annoys me."

"You constantly have to defend yourself and explain and I do not always feel like it. There are people who don't understand me and say 'yes' when it should be 'no'. That annoys me. I rather have them ask me to repeat myself than an answer that makes no sense ... then I withdraw..."

Even the family does not understand

Couples felt misunderstood by their own family. They experienced that relatives with the disease sometimes denied having MD1 and did not want to have anything to do with it. Partners even noted that the parent with MD1 showed little understanding for their own child(ren) with an even more severe form of the disease.

"When it was discovered by her mother, we told everyone to do a DNA test, then you know where you stand. If you see them walking, you can see it. Her own brothers do not know, because they did not do a test." [partner]

From relatives and friends they experienced lack of understanding of what it means to live with MD1. This led to hurtful reactions from others when for instance an appointment had to be cancelled due to ill-health or when they experienced that family members did not make efforts to understand their relative with less intelligible speech.

"When he takes the phone, they always ask for me, because they cannot understand him. And these are his own brothers. Or when they sit next to him, they don't have the patience to let him tell what he wants to say, that sort of things. He talks slowly and it gets worse." [partner]

We manage ourselves

Couples experienced that people are busy with their own lives and forget about them. They preferred to manage themselves and found it difficult to ask for support. Becoming dependent on others or receiving the 'wrong' assistance was a reason for giving up activities and withdrawing.

"We play 'jeu-de-boule' nearby. I cannot pick up a ball, but I have magnets to do that. Then there are people that pick up the ball for me, all with the best intentions, but I hate that so much".

However, when support was given in the 'right' manner, it was highly appreciated. When friends showed understanding and consideration for their restrictions, this was treasured. One participant with MD1 mentioned that their group of friends had asked her to explain about her illness. When she had done so, she experienced better understanding.

> "Last weekend we went to dinner with some good friends. We would have had dinner in a restaurant where the toilet was downstairs, but they then choose a different restaurant. They had thought about that which was fantastic." [partner]

Health care: many islands focused on part of the disease

Participants reported that they often visit many specialists. They felt little attention was paid to the consequences of the disease for their daily life. A couple described the hospital as 'many islands', each looking at one part of the disease. Couples said they visited the hospital together, because that way you hear more. One partner said that she did not mind doing personal care tasks but that she needed proper instructions and support. This couple expressed concerns about coping with a catheter. They wondered whether cycling, attending the choir or swimming was still possible. These questions had not been addressed in the consulting room with healthcare professionals, so they had tried to manage and find solutions themselves.

> "You expect that they tell you these things [how to cope with a catheter] in hospital. Now I need to go to the drugstore. They have something for men who are incontinent. I have thought of it yesterday. You have to do some doctoring yourself. If they had only said how to do it, but they had not." [partner]

Some couples experienced understanding, recognition and support by fellow patients during meetings from the muscular disease association or group medical appointments, a 2 hour medical consultation together with 6 or 7 other patients with the same disease and their partners.

Discussion

This is the first study exploring how couples live and cope with MD1. As such, it offers new insights into the self-management strategies used by couples from the perspective of persons with MD1, their partner and as a couple. Persons with MD1 experienced physical, cognitive and social barriers in daily

life. Their self-management strategies included postponing, avoiding or giving up activities, but also adjusting expectations and finding alternative ways to engage in less demanding activities. Partners reported an increasing burden and felt that if they did not do things, nothing happened. Strategies used by partners included taking over activities, giving directions with written or verbal instructions or prompts. As a couple they faced challenges in finding a mode together with renegotiation of tasks and individual and shared activities. Giving each other freedom, respecting each other's differences in pace and giving scope for their own interests were ways in which couples managed together. Couples experienced lack of understanding from family, friends, society and health care. This lack of understanding contributed to further withdrawal and avoidance of social situations. Couples said they preferred to help each other, rather than asking for help or understanding. Health care was experienced as many islands, with each speciality looking at one part of the disease. Little attention was paid to the consequences of this systemic, neuromuscular disease for their daily life and marital relationship.

Strengths and weaknesses of the study

Although a small, purposive sample (three men and two women with MD1), the strength of the study was that three in depth interviews, lasting between 60-90 minutes, were held with each couple. This resulted in 15 interviews with rich information from three perspectives: persons with MD1, their partners and as a couple. The 1070 codes saturated the four themes and resulted in "theme saturation" as well as "theoretical saturation"[28]. There were no obvious deviant or negative cases, but categories were saturated with a variety of positive and negative experiences. The trustworthiness of the study credibility was promoted through triangulation strategies and other strategies to enhance credibility[25;29].

There was homogeneity in age and the type of MD with the onset of symptoms in adulthood. All participants were Caucasian. There is no information on how MD1 affects the lives of persons from other than western cultures. Younger persons with MD1 might have different experiences, especially since this disease has the characteristic of anticipation, meaning that children of couples with MD1 may have a more severe type of MD revealing problems at an earlier age. The findings give an impression of the lives of middle aged couples with one partner having MD1. However, it is to be expected that there is a considerable group of men with MD1 without a partner. Men with MD1 appear to have a decline in marriage eligibility

whereas women continue to marry at a young age and in a proportion almost equal to that of the unaffected population[30]. In a study of personality patterns of 15 people with MD1, 11 lived alone after divorce or were unmarried[5]. Our research shows the support needs of individuals with MD1 and support strategies provided by their partners. This information may be of value for single persons with MD1 who might be in need of support.

This study also included participants with communication difficulties due to dysarthria in contrast to a previous qualitative study[8]. In some participants intelligibility was limited. The researcher repeated the participants' answers in order to make sure she understood; this repetition also facilitated the transcription.

Although MD1 is the most prevalent adult type of neuromuscular disease, the incidence is approximately 1 in 20.000; these findings may be relevant for couples with other complex chronic illnesses facing physical, cognitive and social impairments which limit everyday activities and social participation.

Meaning of the study: possible explanations and comparison with other studies

Previous studies aiming to get insight in the consequences of MD1 used quantitative approaches[9;31-33], interviewed a heterogeneous group of persons with muscular dystrophies or their partners separately[8;12;34] or focused on living with the hereditary aspect of a muscular disease[13]. This study is the first to combine clients' and partners' perspectives on their daily life experiences and how they cope as couple with MD1.

Lack of understanding of family, friends, society and health care was a major theme which was described in other studies in muscular dystrophy[8;11;12;35;36]. The following factors may be possible explanations: lack of communication, wrong expectations, ignorance and avoidance. Communication was not only an issue because of dysarthria, decreased intelligibility and limited facial expression in persons with MD1. More crucial is the lack of communication about the illness experience of persons living with MD1. How can friends, family and health care professionals understand if they do not communicate about the impact of the disease on daily life? Although patient-centred care is being promoted[37], participants in current study experienced illness–centred care with many health care professionals focusing on part of the disease and its management and not on the consequences for their daily life. A recent review on the management of MD1 confirmed the concentration on impaired functions and structures in medical subsystems[38]. Our study shows that during consultations, doctors (and other

health and social care professionals) would do well to explore the impact of the disease on the couple, rather than focusing on the subsystems of the disease.

Other factors contributing to lack of understanding include wrong expectations and ignorance of other people regarding the potential or limitations of persons with MD1. As MD1 is classified as a neuromuscular disease, muscle weakness and fatigue are expected and treatment is aimed at improving body functions like aerobic capacity, breathing or hand function[14;17;18;39;40]. However, if cognitive problems and social barriers are ignored, efforts to improve physical functions are likely to fail. These barriers may explain why adherence to exercise programs is limited[14]. Current study showed the large impact of cognitive and psychosocial barriers such as lack of initiative and avoidant behaviour for the daily lives of couples. Although these features are well-described characteristics of MD1[5;7;41], they are less obvious and often ignored, leading to wrong interpretations and misjudgements by other people.

From the interviews we have learned that persons with MD1 experienced: 1) being stared at due to their appearance; 2) misunderstanding because of the decreased intelligibility; and 3) being judged as silly or sulky. All of these experiences contributed to avoidance of or withdrawal from social situations. Other social barriers include the inaccessibility of public buildings restricting spontaneous visits to places like museum, cinema or theatre, which was also described by Boström et al.[34]. Although participants tried to hide their disabilities and tended to avoid social situations, they all valued going out and meeting others. Many persons with MD1 experienced difficulties keeping in touch with others leading to feelings of being alone, forlorn and forgotten[8;34]. Breaking down disabling social practices against people with MD1 might be as important, if not more so, than seeking treatment for physical impairments[42]. This is in accordance with Boström and Ahlström who state that: many issues in the management of long-term chronic illness can better be interpreted from a social perspective rather than a biomedical perspective[34].

The self-management strategies of persons with MD1, including adjusting expectations and finding alternative ways to participate with less demands on physical performance, have been described before[34]. These self-management strategies are in contrast with the main rehabilitation interventions which tend to focus on improving physical abilities[14;17;18;39;40]. What can be learned from these self- and couple management strategies is that it is necessary to understand the barriers perceived in daily life, and also

how people adapt their lives to retain their social roles and participate. To support self-management, it is essential to understand the meaning of social participation. Especially when self-management is seen as comprising of three tasks: medical management such as taking medication or doing exercises; role management which involves maintaining, changing and creating meaningful behaviours or life roles, and emotional management including the management of emotions such as fear, frustration and depression, commonly experienced by persons with a chronic disease. [43]. Most health care programmes only deal with medical management. Role-management and emotional management are often being neglected[43]. This study provides insight into how couples themselves fulfilled these different self-management tasks.

Professionals and others primarily focus on the needs of the person with MD1 and overlook the support needs of partners or other family members[12]. Health professionals perceive partners as caregivers rather than care recipients. Also partners do not express their need for support, although they might experience a large caregiver burden, feeling no scope for own interests outside the home which can be a source of despair[12]. This study showed how frustrating and annoying the lack of initiative and avoidant behaviour can be for partners. Previous studies report the difficulties encountered by partners in interpreting whether the need for support is related to MD1 or to lack of will or laziness[12]. Partners are often faced with dilemma's whether to take over and when to encourage their partner with MD1 to do things. Not all partners knew that this so-called lack of will is part of MD1. What can be learned from partners in current study is that knowing that the lack of initiative and avoidant behaviour are related to MD1 helped partners to more consciously apply compensation strategies like prompts or reminders.

Current study showed the resilience of couples to manage together. Supporting each other and giving each other freedom were described as vital couple management strategies. This finding replicates the management strategies used by couples facing other multiple chronic illnesses[44]. Two life philosophies were found: 1) staying positive; and 2) being married means supporting each other whatever happens and coping with what comes along. The combination of individual and shared coping strategies was considered indicative of a healthy balance[44]. A continuum was described between positive support like a helpful reminder and problematic support like nagging or pushing a partner[44]. A balance was promoted in which the person with the disease manages themselves and the partner provides support to

this managing. How assistance is given and received as well as marital interactions that accompany this support, appeared to impact both marital quality and health[44]. These findings support a couple-centred approach for the self-management of couples with complex chronic diseases like MD1 which includes medical management, role management and emotional management for both partners[43].

A couple management approach has shown to be effective for patients with dementia and their caregivers[45;46]. The problems experienced by these couples in relation to coping with loss of abilities, initiative and participation in social activities, decreased quality of life and pressure on family relations and friendships[45] are strikingly similar to the problems experienced in MD1. The consequences of MD1 have been described as comparable to those of the aging population[15]. This is in line with insights that MD1 is considered a progeroid syndrome with accelerated emergence of features of senescence including symptoms of dementia[47]. There is evidence that a community approach aimed at increasing abilities of older people with dementia to engage in meaningful daily occupations and interventions to increase support giving skills of partners increases participation of the person with dementia and reduces caregiver burden[45;46]. Further studies evaluating couple management for couples with other progressive, complex diseases are warranted.

Finally, some participants valued peer support to exchange experiences and learn from fellow patients; whilst others avoided peer support. Boström et al. (2004) stressed the importance of the disability movement with its fellowship for social connections, providing opportunities to meet others and also as an organizer of suitable leisure activities[34]. Another way to meet fellow patients is participation in group consultations with persons having MD1, their partners and their physician in which the topics are often chosen by the participants. During such visits, participants learn from each other and physicians learn from the experiences of patients and their partners[48]. This approach has been promoted as a way for medical students to gain understanding of what it is like to live with a chronic disease and how couples adjust and cope[48].

Implications for clinicians and policymakers

Although there is mounting evidence for effective system changes that improve chronic care, such as the Chronic Care Model[49-51], health care often remains with basic structures and practices designed for acute diseases[48]. The traditional medical care model mainly focuses on treatment of

impairments[6], ignoring the illness experience and its impact on the system, including partners. Our recommendations for clinicians therefore include:

1. A shift from the focus on physical functions to a person-, or even a couple-centred approach aiming to understand not only the disease, but also the illness experience of persons with a complex chronic illness and their partners. This shift involves:
- A more narrative approach besides the traditional biomedical analytical approach; and
- Referral of patients and their partners to appropriate interdisciplinary medical, rehabilitation and community services to address barriers from medical as well as psychological and social perspectives.
2. A shift from self-management to couple management in complex chronic conditions, in which self-management skills of both partners are supported in order to:
- Maximize participation for persons with a complex chronic disease such as MD1;
- Reduce caregiver burden by enhancing support giving skills;
- Find a healthy balance together in shared and individual activities.

This study showed how important it is that clinicians use an integrated medical and social approach for persons with MD1 in which couple management is the starting point.

Future research

Challenges for the future include further development and evaluation of the acceptability and effectiveness of a couple-centred approach for complex chronic illnesses. A couple-centred approach includes the three aspects of self-management: medical management, role management and emotional management[43] and should also address disabling social practices. As this can be considered a complex intervention including several components[52;53], it is recommended to use a stepwise approach as described in the framework for design and evaluation of complex interventions[52].

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Chapter 8
General discussion

Part I: Professional perspective

Part I of this thesis was based on the premise that utilization of physical therapy (PT), occupational therapy (OT) and speech therapy (ST) in persons with neuromuscular diseases (NMD) was inefficient. It was hypothesized that a multidisciplinary approach with single consultations and advice would result in more efficient referral to and utilization of OT, PT and ST for persons with NMD. In the absence of evidence based guidelines, the content of the consultations and advices was merely based on professional opinion from the OT, PT and ST from the neuromuscular expert centre. These advices were sent to colleagues in primary care and rehabilitation settings. Utilization of OT, PT and ST based on these advices was compared with usual care regarding volume of therapy and costs in *Chapter 2*. Implementation of the advices and possible barriers for implementation were explored in *Chapter 3*.

Overuse and underuse of allied health care

According to our comparative cohort study, in current practice there was overuse as well as underuse regarding PT and underuse regarding OT and ST. Compared to current practice, the PT advices suggested more short periods (boosts) of PT addressing and evaluating specific goals and supporting self-management regarding physical exercises in daily life. The advices also suggested more short periods of OT compared to current practice with goals aimed at enabling valued occupations by means of giving instruction and advice regarding aids and adaptations or education regarding energy conservation strategies. For ST, 40% of the persons received advice during single consultations, which might include slowing down speaking rate to improve intelligibility or postural and dietary advice to improve eating and drinking abilities.

The rationale behind the advices was to increase self-management of persons with NMD and to become a coach for persons with NMD instead of providing ongoing therapy. It was expected that such approach would improve efficient utilization of PT, OT and ST for persons with NMD and to decrease health care costs. This approach is in agreement with current views on the management of chronic diseases[1-4], in which self-management is promoted. Self-management support however requires changes in professional thinking and behavior[3]. It also requires a change for persons with a chronic disease who are encouraged to gain the information, skills and confidence to manage and live with their condition[4].

In current practice less than 10% of the persons with NMD had OT whereas there appeared to be an indication for short term OT in 45% of the persons assessed. This difference is most likely due to lack of knowledge and insight of what OT has to offer for persons with NMD. The lack of evidence for OT in NMD adds to this lack of knowledge, which calls for actions to increase the evidence base and provide insight into the benefits of OT to support persons with NMD in their abilities to carry out valued tasks and life roles.

In current practice 70% of the PTs saw their patients with NMD at least once weekly for a year or more. Despite lack of scientific evidence to support this need for ongoing PT, advice to reduce the volume of PT was implemented in only 15% of the cases. Many different factors may have contributed to this, including the difficulties persons with NMD may experience difficulties in implementing physical exercise in daily life without support of a therapist and outside the therapy practice.

Persons with NMD hardly make use of ST services, although a considerable number of persons has problems with communication or swallowing. Although the impairments cannot be cured, compensatory strategies are explained and instructions given often withinin a single consultation. Compared to the many advices for short term PT and OT interventions, further ST interventions were advised in only a very limited number of persons with NMD.

Implementation of the advices and possible barriers for implementation

Our comparative cohort study concluded that there was limited implementation of the advices. In this study the amount of therapy before and following the advices was compared. Further analyses showed a different picture. Implementation of the OT advices in primary care was excellent (100%, n=21), whereas the OT advices (n=10) referred to a rehabilitation physician were implemented in 58% of the persons. There were various reasons for this lack of implementation and these reasons were at the level of the advice, the patient, the professional and the organization.

Implementation of PT advice was also excellent (100%, n= 58) when it did not involve a change in volume of PT. When the advice suggested a change in volume (reduction, discontinuation or initiation of PT), the advice was implemented in 25% (7 of 28 persons). Possible factors influencing implementation were found at the level of the advice, the patient, the professional and the organization of practice. This showed that we are dealing with complex interventions comprising many different components.

To gain insight into components from the perspective of the professionals, follow-up telephone calls were recommended between colleagues from the neuromuscular centre and colleagues in community or rehabilitation setting to discuss the advice and possible barriers for implementation.

Insights from the perspective of quality of health care improvements have indicated that implementing changes requires a whole systems approach engaging the patient, professionals and service organization[3]. An example of a change is when persons with NMD are encouraged to do physical exercises or sports in daily life themselves without the ongoing support of a therapist. Studies from the perspective of persons with chronic neurological conditions and from the perspective of PTs showed that many different barriers and facilitators are involved regarding participation in physical activities and exercise or sport in a public facility[5;6]. The presence of professionals that are knowledgeable about the neurological conditions is perceived as a facilitator, whereas a sports or exercise facility without such staff is perceived as a barrier for participation. Other barriers included accessibility of facilities, transport, costs or fees and feelings of shame or discomfort to sport among healthy persons. These findings support the complexity of various components involved when the aim is to support self-management in for instance physical activities and exercise. The costs involved in medical fitness in the Netherlands generally are for the persons themselves, even when there is a medical indication. Individual exercise therapy by a PT however, is often fully reimbursed by the health insurances. This might have been one of the components influencing implementation of advice to reduce PT and instead participate in physical exercise in for instance medical fitness.

Limitations

Our studies on professional advice from a neuromuscular centre had several limitations. At first, the use of different types of questionnaires regarding therapy volume from different perspectives resulting in different outcomes; In the first study comparing usual practice with allied health care advices from a neuromuscular centre, we used data from the patient health care diaries and patient questionnaires at six months follow up. In the research on implementation of the advice, we also used professional questionnaires at six months follow up. This sometimes resulted in contradictory information showing the presence of recall bias. At six months follow up some patients

recalled not to have had OT, whereas they had had OT based on the professional questionnaires.

Another reason for different results regarding implementation between the two chapters (*2 and 3*) was related to the criteria used for successful implementation. In *Chapter 2*, the number of treatment sessions advised was compared with the number of treatment sessions recorded by patients. In *Chapter 3* implementation was considered successful for OT when persons who had been advised OT, actually had received OT, regardless the number of therapy sessions. For PT, five categories were used (continue same volume, continue no PT, reduce volume, initiate volume or discontinue PT altogether) to determine whether implementation had been successful. This showed a different picture of the extent of implementation in *Chapter 3*.

Another limitation of the study involved the lack of patient oriented outcomes to explore the effectiveness of the multidisciplinary advice on health and well-being of persons with NMD. Current exploratory study focused solely on allied health utilization and implementation of the suggested volume of therapy. If future studies aim to measure patient-oriented outcomes, further research is needed regarding the most appropriate measures at the level of body functions, activities and participation, personal and contextual factors as well as registration of the content of therapies and the volume of therapy provided to be able to explore the outcome thereof.

Lack of scientific evidence to support the treatment advices was also experienced as a limitation and showed the need for additional literature searches *(Chapters 4, 5 and 6)* and studies to be undertaken in the field. And finally, it was felt that single consultations provided limited insight into the daily lives of persons with NMD in the community and how they managed to live with the disease. Although the OT, PT and ST professionals aimed to apply a client-centered approach, by asking the persons to tell about the impact of the disease for their daily lives, it was felt that the client perspective could not be fully understood in the restricted time allocated for the consultations which took place in the clinical context of a hospital. To address this, a qualitative study was conducted aiming to gain in-depth insight and understanding of how persons with myotonic dystrophy type 1 (MD1), the most prevalent neuromuscular disease among adults and their partners experience living with this disease and how they manage themselves in their own context *(Chapter 7)*.

Part II: The scientific perspective

To address the problem of limited scientific evidence for OT, PT and ST for persons with NMD, three systematic reviews were conducted (*Chapters 4, 5 and 6*). These reviews not only included evidence from RCTs and CCTs, but also from uncontrolled pre-post designs. The findings from the systematic reviews have shown that the evidence base for OT and ST for persons with NMD is weak and based on only one (ST) or two (OT) small uncontrolled studies[7;8]. The highest level of evidence (level II evidence) was found for PT for persons with NMD, specifically for aerobic exercise in combination with strengthening exercises[9].

Occupational therapy

For OT, there are indications that training of hand function is effective in persons with Welander muscular dystrophy (n=12)[10] and myotonic dystrophy (MD) (n=5)[11]. The first diagnosis is extremely rare in the Netherlands with only a few people having the disease[12]. MD on the other hand is one of the most prevalent NMD[13]. The evidence from the study with persons having MD has already been applied in neuromuscular teams in the Netherlands, which has resulted in a description of two cases, showing improvements in strength and ability to use the hand in daily occupations [14]. These findings also resulted in increased advice for training of hand function in persons with MD who had been admitted in the neuromuscular expertise and consultation centre in Nijmegen. Previously, interventions were merely aimed at compensation for the loss of hand function instead of training. The training studies have shown that there are still possibilities to train and improve hand function in persons with MD. However, our own experiences and results applying this hand function training in practice varied. Some were in agreement with the positive research findings and case descriptions, but others were not, which was due to difficulties with adherence to such program. Problems with adherence is a well-known problem in rehabilitation of persons with MD1[14;15]. In practice, additional monitoring and encouragement is often needed. Evidence for long-term efficacy of this intervention is still lacking.

Currently, most OT interventions in NMD include advice and training in adaptive strategies to compensate for the loss of functions and to promote participation in activities and life roles. These adaptive strategies involve the use of aids and adaptations, the use of compensatory movements or postures, and simplifying or eliminating tasks to save energy for participation in

meaningful activities. Although these interventions have not been the focus in studies on OT for persons with NMD, studies in other chronic conditions have shown the effectiveness of OT. For instance, energy conservation strategies have been effective in MS to decrease the impact of fatigue[16;17]. Severe fatigue is also present in the majority (61-74%) of patients with NMD[18]. Energy conservation strategies are therefore regularly advised in OT practice in persons with NMD[19] and research in this area is recommended.

Other fields providing evidence for the efficacy of OT include interventions aimed at decreasing the incidence of falls in elderly people living at home with high risk of falling[20]. The interventions included advice on assistive devices as part of a home hazards assessment or training of skills combined with a home hazard assessment. Because falling is a clinically important problem in NMD [21], such interventions might also be efficacious for the treatment of persons with NMD at risk of falling.

There is also evidence for the efficacy of comprehensive OT on functional ability, social participation and quality of life in community dwelling elderly[20]. Community based OT has shown to be effective in improving daily functioning of older people with dementia and to reduce care giver burden[22-24]. As some neuromuscular diseases like myotonic dystrophy are considered progeroid syndromes with accelerated emergence of features of senescence including symptoms of dementia[25] evidence for the effectiveness of OT in the elderly with dementia may be applicable to some persons with NMD. And although clinically there is no debate regarding the benefits of OT in NMD, further studies are needed to provide OT in NMD with a firm scientific base.

Physical therapy

Regarding PT, aerobic exercise in combination with strengthening exercises is likely to be effective for persons with muscle diseases (level II evidence). There are indications that it is also effective in a heterogeneous group of NMD (level III evidence). Besides, level III evidence was found for aerobic exercises in persons with muscle disorder and for breathing exercises for persons with myasthenia gravis (MG) and MD. Adverse effects were negligible.

This level II evidence for the combination of muscle strengthening and aerobic exercises found in current review was discarded in a Cochrane review on strength training and aerobic exercise training for muscle disease [26]. Reason was the exclusion of training programs with duration of less than

10 weeks or studies that were not randomised. The only study included in the Cochrane review was the low quality RCT resulting in the conclusion that aerobic exercise in combination with strength training appears to be safe and may be effective in increasing submaximal endurance capacity in people with mitochondrial myopathy. This Cochrane review concluded that more research is needed before exercise programmes are generally prescribed in muscle diseases[26].

Another Cochrane review on Rehabilitation interventions for foot drop in NMD[27] included an RCT which was also included in the review in current thesis. In this RCT[28] strength training had a significant beneficial effect on walking ability in people with Charcot-Marie-Tooth disease (hereditary motor and sensory neuropathy, HMSN), but no significant effect on walking ability in people with MD.

The conclusions in both Cochrane reviews were based on studies also included in the review in current thesis. However, the conclusions differed due to other in- and exclusion criteria of studies or different criteria for providing evidence for effectiveness. The review in current thesis considered studies to provide evidence when at least half of the variables in the domain body functions or activities/participation showed a significant effect instead of only one out of six, which was the case in the study on strength training in HMSN[27;28]. The use of primary outcome measures is recommended instead of using many different outcomes, which had been the case in most studies in our systematic review.

Two more recent Cochrane reviews in the field of PT in NMD were found. One reported no evidence for interventions for increasing ankle range of motion in Charcot-Marie-Tooth disease type 1A or Duchenne muscular dystrophy[29]. Another Cochrane review on therapeutic exercise for people with amyotrophic lateral sclerosis (ALS) or motor neuron disease concluded that the studies detected were too small to determine to what extent strengthening exercises for people with ALS are beneficial, or whether exercise is harmful[30].

Other reviews focusing on exercise therapy in adult persons with inflammatory myopathies, concluded that exercise therapy in adults with polymyositis and dermatomyositis was safe and efficacious and can be recommended [31;32]. Regarding the combination of strengthening and aerobic exercises, this conclusion was in agreement with the findings from our review in this thesis. Regarding muscle strengthening exercises, the conclusions differed. This also could be explained by differences in studies

included and in criteria for effectiveness. Remarkable was the presence of adverse effects in a recent study in two of eight participants (increased joint swelling and tenderness and diffuse tendinitis pain influencing exercise capacity)[33]. However, in the conclusions this study emphasized lack of muscle inflammation based on muscle biopsies. The absence of adverse effects so far legitimated the use of exercise therapy. This study showed that although exercise therapy did not show muscle inflammation, other adverse effects were present.

At the time of the review, there were no studies on interventions to improve mobility including transfers and walking. A recent study evaluated posture and gait abilities in patients with MD following a rehabilitation program[34]. However, this retrospective analysis of medical files of 20 patients, showing improvements in five of six outcome measures at the level of strength and one of six outcome measures at the level of gait, would have been discarded due to lack of methodological quality.

In conclusion, lack of uniformity in type of interventions, intensity of exercise therapy and type of outcome measures hamper comparison between studies. Clearly more uniformity is recommended. Also research designs other than RCTs and CCTs, such as observational studies, should be recognized as very valuable alternatives to provide evidence for the course of PT interventions and the possible effectiveness in persons with NMD.

Speech therapy

For ST, there were indications for the efficacy of correcting head position in persons with OPMD[8;35]. In clinical practice, many persons with an NMD other than OPMD have ptosis. Instructions regarding the influence of posture of head and neck on swallowing might therefore be applicable to many persons with NMD, particularly when the disease leads to changes in posture or weakness in the head-neck-shoulder region or when the disease leads to weakness in muscles involved in speech or swallowing. This was confirmed in a study showing better swallowing in persons with spinal muscular atrophy (SMA) type II, when the head was positioned more forward[36].

Dysphagia and dysarthria diagnosed and treated by ST, are relatively prevalent and clinically relevant symptoms in persons with NMD. Dysarthria negatively influences communication and social participation, it has large impact on quality of life of persons[37]. The aim of ST interventions is to improve the intelligibility of persons with dysarthria due to NMD. Even if the dysarthria itself continues to deteriorate, compensational strategies such as

slowing speaking rate, speaking face to face, using alternative words, concentration on speaking only and energy conservation may prolong the time of successful communication [37]. Studies have shown that it is possible for persons with amyotrophic lateral sclerosis (ALS) to alter speaking rate, although there is lack of evidence that it has effect on intelligibility[38;39].

Van Nuffelen et al.[39] recently have studied seven rate control methods on speech intelligibility, speaking rate, and articulation rate and pause characteristics in various types and severity of dysarthria. They found that each rate control method decreased both speaking rate and articulation rate, although the decrease was not significant for the strategy 'speaking slower on demand'. The results showed that each rate control method had the potential to establish a significant improvement in speech intelligibility for various types of dysarthria. The most effective methods were hand tapping, pacing board and alphabet board. It was therefore recommended that clinicians judge the effect of rate control for an individual with dysarthria during a trial session, trying out different methods at different rates. However, they also mentioned that although rate control may improve intelligibility in dysarthria, it may also have the opposite effect on intelligibility in some cases[39].

Obviously, there is a need for studies to research the effectiveness of ST advice on successful communication. This cannot be assessed by measuring speech intelligibility in a clinic room, but warrants assessment in the individual's actual life situations. It is a challenge to use assessments that represent the perspective of the persons experiencing problems in communication, who is ultimately the best person to judge whether participation is adequate and successful[40]. A Communicative Participation Item Bank is being developed, rating the extent to which a condition interferes with participation in various speaking communication situations[41].

Although the ultimate goal of ST is to improve communication and eating and drinking in daily life, which is the level of activities and participation in terms of the International Classification of Functioning, Disability and Health (ICF)[42], assessment and management are also aimed at the level of body functions influencing communication and eating and drinking. A recent study on lip strengthening exercises in children and adolescents (7-19 years) with MD1, provided indications for possible effectiveness on maximal lip force and lip force endurance in a group of persons having moderate to severe impairments in lip force[43]. It is suggested that lip strengthening exercises can be a complement to other ST

interventions to improve articulation, saliva control, or eating and drinking ability, all of which are problematic in MD1[44]. It should be noted however that these findings are based on studies with children. The effectiveness of such interventions in adults may not have the same effect.

Other ST interventions such as the provision of information and advice on dietary modification and augmentative and alternative communication have not been studied in persons with NMD. Further research in this area is therefore recommended.

Part III: Client perspective

Our qualitative study showed that health care for persons with MD1 is mainly focused on the disease and body functions, rather than on the persons having the disease. There is lack of understanding of the experiences and consequences of MD1 for the lives of persons with the disease and their partners. In addition to the most obvious barriers of reduced mobility, poor physical fitness and fatigue, persons with MD1 experience several other disabling barriers which are less obvious and not always adequately addressed. These include the cognitive barriers, psychosocial barriers, and societal barriers. Addressing these barriers and breaking down disabling social practices against people with MD1 might be as important, if not more so, than seeking treatment for physical impairments[45;46]. Here lies a challenge for OT, PT and ST for MD1 in providing support to overcome the different types of barriers perceived.

Another important finding from our qualitative study is the large impact of a complex chronic disorder like MD1 on the lives of partners and on the lives of both as a couple. Partners may experience an enormous burden as the decrease in abilities and initiative of the person with MD1 leads to an increase in demands, tasks and responsibilities of partners. They described this as a gradual and natural process, but also as frustrating and annoying experiences. Due to the differences in interests, physical and cognitive abilities, couples regularly have to renegotiate tasks and roles and find a balance between shared and individual activities. So far health care is mainly focused of persons with the disease. Our findings however, suggest a couple-centred approach aiming to maximize participation for persons with MD1, reduce caregiver burden by enhancing support giving skills, and find a healthy balance together in shared and individual activities. Such couple management approach has shown to be effective in couples living in the community and in which one of the partners has dementia[22-24].

Traditionally the focus of health care is on medical management of a condition, whereas self-management of a chronic condition like MD1 comprises of two other tasks: role management (maintaining, changing, and creating new meaningful behaviors or life roles) and emotional management (learning to manage emotions such as anger, fear, frustration, and depression) [47]. Role-management and emotional management have often been neglected in health care and here lies a role for allied health professionals including OT, PT and ST as part of a coordinated multidisciplinary team providing appropriate services and support when needed. To support persons in these types of self-management, it is essential to understand the meaning of social participation for persons, the barriers perceived and how persons adapt their lives to retain their social roles and participation.

In client-centered practice, the expertise of clients is considered as important as the expertise of professionals and in practice they share this knowledge and share decision making. This partnership paradigm implies that while professionals are experts about diseases, patients are experts about their own lives. Our study showed how persons with MD1 and their partners manage their lives together, which involved adjusting expectations and participation in activities and roles with less demands on physical performance and renegotiation of (marital) roles (role management). In this process persons with MD1 and their partners had to cope with frustrations, dilemmas and caregiver burden (emotional management).

A client-centred approach or the partnership paradigm is increasingly being implemented in health care for chronically ill persons. Moreover, also in research the client-perspective is increasingly being incorporated. This has led to priority setting in research from the perspective of persons with NMD[48]. Four main research themes were distinguished as well as priorities within these themes: 1) Health status with emphasis on finding a cure or prevention of deterioration. This includes research into fatigue, pain and other specific complaints; 2) Quality of life with emphasis on partner relation, family life and social contacts. Also mobility, work education and leisure were topics considered important research topics; 3) Quality of health care with emphasis on mobility and training followed by aids and adaptations and professional support; 4) General issues with the main issue of independence and other issues like understanding, information provision[48].

Perspective of effective service delivery: Chronic Care Model

Studies in current thesis have presented the state of the art regarding the components of evidence based OT, PT and ST for persons with NMD. There are still gaps to bridge regarding: 1) professional expertise; 2) scientific evidence; 3) patient-centered care, including next of kin. In identifying areas for development to bridge these gaps, the Chronic Care Model (CCM) appeared useful[4]. The key-elements of evidence based practice in the context (Figure 1) can be compared with the key-elements of the CCM (Figure 2).

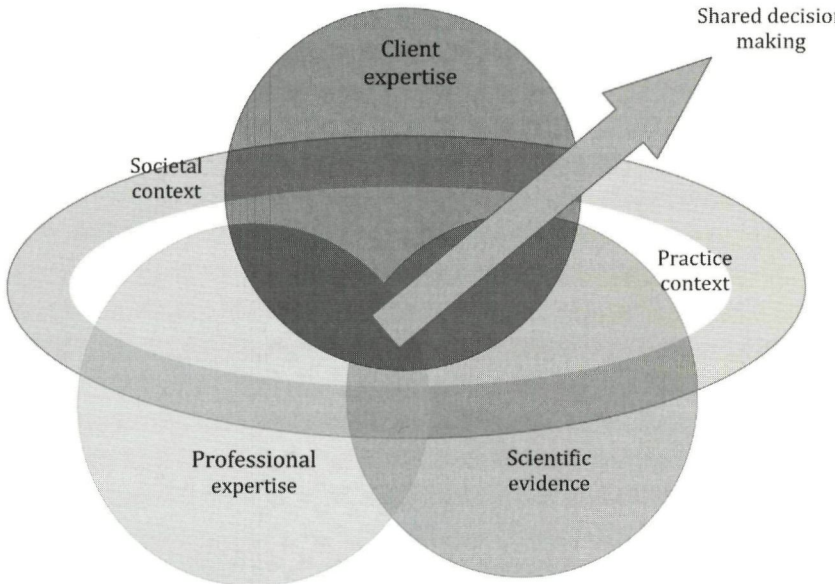

Figure 1. Elements of evidence based practice (EBP) in a practice context and societal context.

In the CCM the professional expertise is congruent with a proactive prepared practice team and the client expertise with the informed activated patient (and families). Together they have productive interactions (including shared decision making) leading to improved outcomes. To support and enable these productive interactions, developments in the health care system are promoted, which include self-management support, delivery system design, decision support and clinical information systems. Self-management support is provided within health care context and in community.

The CCM has its roots in evidence based medicine and is a visual compilation of evidence about the determinants of effective chronic illness care and the nature of practice systems that facilitate good care and good patient outcomes[4]. Therefore, elements of CCM may assist in improving efficient service delivery of OT, PT and ST in persons with NMD. Specifically, the elements self-management support, delivery system design, decision support, clinical information systems and outcomes are areas for development and improvement.

Self-management support

Self-management support involves assisting and coaching persons with a chronic condition and their families to acquire skills and confidence to manage the challenges of living with a chronic condition like NMD. In the past, persons with a chronic condition passively received treatment to restore or maintain body functions. Self-management support aims to activate persons to be informed and able to manage their condition themselves. For persons with a chronic condition and for professionals such as OT, PT and ST, this requires a different way of thinking and working requiring different skills like motivational interviewing to support patients in their self-management[3;49].

Self-management support requires a systems approach including the patient, the professionals and service organisation (health care and community resources)[3]. An example of a self-management approach is the Project Shake-It-Up, promoting health and empowerment of persons with different neurological disability and also building the capacity of community organisations that provide services to this population[45;46]. Other initiatives include our own multidisciplinary program for peers with chronic neurological conditions such as NMD[50]. This program includes several aspects of self-management support: physical training and education on training and on energy conservation strategies[50] as well as support to enable participation in sports or physical activities at home and in community. This program is currently being piloted. Further development and testing of these self-management programs is needed for which the use of the framework of developing and testing complex interventions is recommended[51].

Delivery system design

As visualized in the CCM, self-management support is on the edge of health care system and community resources. This is where professionals from

health care settings and community resources collaborate. This can be within the same profession, but also between professionals and organizations. The use of internet communities may provide opportunities to share experiences and discuss treatment options[52]. Expert teams in specialized centers are recommended for diagnostic purposes and treatment advice, whereas for continuity of care, support in the home situation and in community is recommended with respect for the social context and autonomy of the persons with chronic illness[49]. This is in agreement with the promotion of an integrated multidisciplinary approach for the management of NMD involving all members of the patients' environment including family, clinicians, decision-makers and community organizations to move out of the spiral of disease and handicap and toward optimal citizenship and quality of life[15;53;54].

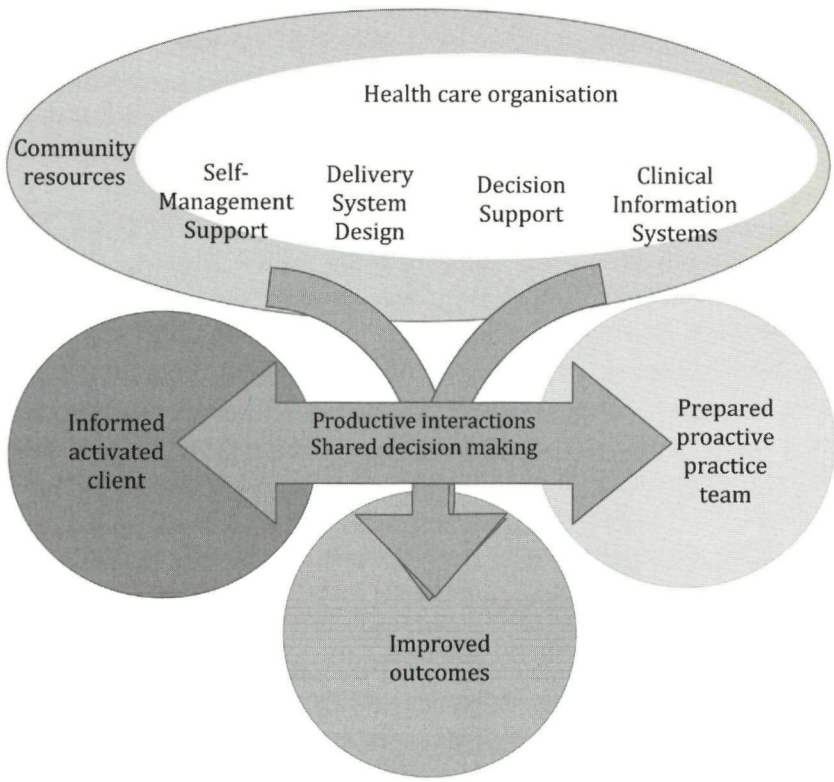

Figure 2. Elements of chronic care model (CCM) by Wagner et al.[4].

Decision support and clinical information systems

When persons with NMD are admitted to the neuromuscular centre for diagnostic purposes and treatment advice, they complete a questionnaire regarding perceived problems in activities and participation and the need for support in different areas (PLANQ)[55-57]. The PLAN-Q is developed to be used as a decision support to guide referral to OT, PT and ST. The questionnaire also provides valuable information for the preparation of the consultations. Reports from the consultations are registered in a database accessible for members of the team. During a multidisciplinary meeting the findings are discussed within the team and results in advice for medical and rehabilitation interventions.

Such standardized and organized approaches to collect, summarize and review individual or aggregate patient data are recommended to facilitate care[4]. A computerized disease registry that includes critical information about each patient and results of important aspects of care enable care teams to call in patients with specific needs, deliver planned care, receive feedback, and implement reminder systems[4]. Such systems will also assist in the development of clinical guidelines and will provide information which can lead to practice based evidence.

Outcomes

When evaluating OT, PT and ST interventions and also to guide referral to OT, PT and ST, valid outcome measures are required. From the perspective of persons with NMD[48] the following themes need to be addressed and assessed using different levels of the International Classification of Functioning, Disability and Health (ICF)[42]:

1) At the level of body functions and general health condition: muscle strength, fatigue, pain and other specific complaints such as balance problems;

2) At the level of activities, participation and quality of life: mobility, work, education, leisure, partner relation, family life and social contacts;

3) At the level of personal and environmental factors: self-efficacy, coping, provision of support, information, aids and adaptations and psychosocial support.

Further systematic reviews and/or consensus meetings with researchers, professionals and patients with NMD are recommended to agree on outcome measures for clinical and research purposes regarding the rehabilitation of persons with NMD.

Conclusions and recommendations for future service delivery and research

To promote efficient service delivery of OT, PT and ST for persons with NMD at the level of increasing professional expertise, building the scientific evidence base and using client expertise, recommendations regarding future service delivery and research include:

- Exchange of knowledge and experience with the use of on-line internet communities, which will increase expertise of professionals in neuromuscular centers as well as professionals in community;
- Observational studies with the use of clinical information registries in which patient data are systematically recorded. A practice based evidence approach is recommended[58-60] collecting comprehensive patient, treatment and outcome data to uncover best practices, which can be subject for further research;
- For this purpose there is a need for a selection of standardized and individualized patient–oriented outcome measures at the level of diseases, body functions and structure, activities and participation, personal factors and contextual factors to be administered by patients and professionals before and during consultations using questionnaires, interviews, tests and observations;
- Development and implementation of decision support tools such as referral guidelines[55-57] and clinical guidelines for OT, PT and ST in different types of NMD;
- Development, testing and implementing self-management programs aiming at supporting persons with NMD and their next of kin in meaningful social participation. To achieve this, the use of the MRC framework for developing and testing complex interventions is recommended[51].

In conclusion, it is suggested to make use of practice informing research as well as research informing practice[60] using different qualitative and quantitative research designs to increase the evidence base for OT, PT and ST in persons with NMD.

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Chapter 9
Summary

In this thesis the state of the art regarding occupational therapy (OT), physical therapy (PT) and speech therapy (ST) for persons with neuromuscular diseases (NMD) is presented from three perspectives: 1) professional perspective; 2) scientific perspective; and 3) client perspective. These perspectives comprise the elements of evidence based practice and clinical decision making (*Chapter 1*).

Current allied health care (OT, PT and ST) for persons with NMD is characterized by large variation in referral, content, duration, and knowledge among referring physicians, professionals, and clients. This thesis aims to increase and integrate knowledge and insights from the perspectives above regarding OT, PT, and ST for persons with NMD.

Part I of this thesis represents the **professional perspective**. In a cohort study the volume of OT, PT and ST as currently received by persons with NMD (reference group n=106) was compared to the volume of OT, PT and ST based on an integrated advice recommended by a multidisciplinary team from a neuromuscular expert centre (intervention group n=102). These advices were composed following single OT, PT and ST consultations and a multidisciplinary meeting. The volume (frequency and duration) of OT, PT and ST for the reference group was studied retrospectively and prospectively with the use of patient questionnaires at baseline and a health care diary during 8 weeks follow up. In the intervention group, questionnaires were completed by patients and professionals (OT, PT, and ST or rehabilitation physician) at baseline, following the consultations and advice, and at 6 months follow up.

In a cost analysis (*Chapter 2*), outcome variables included volume of therapy, relative over- and underuse of therapy, and costs of therapy and intervention (consults and multidisciplinary meeting resulting in advice). When current service utilization was compared to the multidisciplinary advice given, there was 40% underuse of OT among persons with NMD. For PT, there was 32% overuse and 22% underuse; for ST, there was neither over- nor underuse. Forty percent of persons with NMD received ST advice during single consultations with no indication for further ST interventions. For OT, 27% received advice during single consultations and for PT this was the case for 19% of the persons with NMD. If fully implemented, the multidisciplinary approach would result in a mean cost savings of € 85,- per person (costs of the multidisciplinary advice €245,- excluded). The recommended therapy had, however, been implemented only partially at 6

months follow up. It is concluded that some persons with a NMD did not receive any form of OT, PT or ST, where they might need it. Among persons with NMD who received allied health care, quite a few received these treatments for too long periods of time. Ways need to be developed to improve implementation of the multidisciplinary advice to obtain a more favorable balance between its costs and benefits.

At 6 months follow up, implementation of the advice was evaluated for OT, PT and ST and possible barriers to implementation were explored at the level of the advice itself, the person with NMD, the professional or the health care organization *(Chapter 3)*. The criterion for successful implementation for OT and ST was defined as having had therapy when advised so. This was evaluated for advice sent to primary care and to rehabilitation setting separately. For PT we used different criteria for implementation because a considerable number of persons with NMD already had PT. Five categories for advice were distinguished: continue *not* to have PT, discontinue PT, reduce the amount of PT, continue the same amount of PT, or initiate PT. For each category, the percentage in which the advice was successfully implemented was calculated.

Results showed that advice for OT and ST was fully implemented in primary care. However, advice for OT was only partially (58%) implemented in a rehabilitation setting. For PT, advice to continue PT (n=64) was almost fully implemented. In contrast, implementation of advice recommending a change in the amount of PT was significantly less ($p<0.001$). Advice to reduce the amount of PT was implemented in 15%, advice to discontinue PT was implemented in 37.5%, and advice to initiate PT was implemented in 29% of the persons with NMD. Possible barriers for implementation were related to the advice itself (feasibility of treatment duration, correctness and completeness), the person with NMD (motivation), and the professional (experience in treatment of NMDs). Therapists expressed a desire to discuss the treatment advice with the multidisciplinary team. Follow-up telephone calls have been recommended to provide therapists an opportunity for discussion.

Part II represents the **scientific perspective** with three systematic reviews on respectively the scientific evidence for OT, PT and ST for adults with NMD. Comprehensive searches were performed for controlled and uncontrolled studies in the Cochrane Library, Medline, CINAHL and Embase databases. Search strategies included a search for different types of NMD, a search for

different research designs in combination with searches for OT (*Chapter 4*), PT *(Chapter 5)* and ST *(Chapter 6).* A best-evidence synthesis was performed using three levels of evidence. Level I evidence refers to at least 2 RCTs (Randomized Controlled Trials) of sufficient quality. Level II evidence refers to 1 good quality RCT or at least 2 independent controlled studies (RCTs or Controlled Clinical Trials, CCTs) of less methodological quality, and level III evidence refers to an RCT or CCT of low methodological quality or at least 1 other design of sufficient methodological quality. When inconsistent findings were found in studies of similar design and methodological quality, conclusions were formulated as 'there is insufficient evidence that....'.

For OT, only two other designs were found which provided level III evidence for the efficacy of hand training in muscle diseases. There is a lack of scientific evidence regarding the question whether, through occupational therapy, persons with NMD are better capable of participating in meaningful activities and roles. For PT, there was level II evidence for aerobic exercises in combination with strengthening exercises for persons with muscle disorders. There was level III evidence that aerobic exercises with or without strengthening exercises were also effective in other NMD. Other studies provided level III evidence for the efficacy of breathing exercises in persons with myasthenia and myotonic dystrophy. For ST, only one study provided level III evidence that correction of head position in persons with oculopharyngeal muscular dystrophy improves swallowing efficiency.

Part III represents the **client perspective.** A qualitative study was carried out with five couples, including three men and two women with myotonic dystrophy type 1 (MD1) and their partners (*Chapter 7).* The aim was to increase understanding of how MD1 affects the lives of couples and how they themselves manage individually and together to better match health care to their needs. Each couple was visited twice at home. During the first visit individual in-depth interviews were held, and during the second visit a couple interview was carried out. Findings were described from the perspective of persons with MD1, their partners and from both as a couple.

People with MD1 associate this progressive, neuromuscular condition with decreasing abilities, describing physical, cognitive and psychosocial barriers to everyday activities and social participation. Partners highlighted the increasing burden of care giving, giving directions and using reminders to compensate for the lack of initiative and avoidant behaviour due to MD1. Couples portrayed the dilemmas and frustrations of renegotiating marital

roles and responsibilities; stressing the importance of achieving a balance between individual and shared activities. All participants experienced a lack of understanding from relatives, friends, and society, leading to withdrawal and isolation. Health care was perceived as fragmentary, with specialists focusing on specific parts of the body rather than seeking to understand the implications of the systemic disorder on daily life.

This study indicates that people with MD1 and their partners experience a host of problems, some of which are unexpected, underestimated and neglected, such as the cognitive problems and social barriers, which affect their quality of life and relationship. Couples felt misunderstood, perceiving deterioration as inevitable and that nothing could be done. Learning from these five couples has produced some practical recommendations that challenge the tendency to treat MD1 as a condition with primarily physical impairments. It is vital to listen to couples, to elicit the impact of MD1, as a multisystem disorder that influences every aspect of their life together. Couple management, supporting the self-management skills of both partners is proposed as a way of reducing the mismatch between health services and health needs.

In *Chapter 8* all findings are discussed and the Chronic Care Model (CCM) is introduced as a useful model, comprising of elements to improve efficient service delivery of OT, PT and ST in persons with NMD. Specifically, the elements self-management support, delivery system design, decision support, clinical information systems and outcomes are areas for development and improvement.

Conclusions and recommendations

Current service delivery of OT, PT and ST for persons with NMD is not efficient and advice from a neuromuscular team to increase the efficiency is partially implemented. Besides, the evidence base for OT, PT and ST is limited. Finally, couples with MD1 experience far more problems in daily life than are addressed in health care. Therefore, recommendations regarding future service delivery and research include:

- Further exchange of knowledge and experience among professionals;
- Use of standardized and patient–oriented outcome measures at the level of diseases, body functions and structure, activities and participation, personal factors and contextual factors to be administered by patients and professionals;

- Development and implementation of decision support tools such as referral guidelines and clinical guidelines for OT, PT and ST in different types of NMD;
- Performance of observational studies with the use of clinical information registries in which patient data are systematically recorded;
- Development, testing and implementing self-management programs aiming at supporting persons with NMD and their next of kin in meaningful social participation; to achieve this, the use of the Medical Research Council framework for developing and testing complex interventions is recommended.

Chapter 10
Samenvatting

In dit proefschrift wordt de 'state of the art' ten aanzien van ergotherapie (ET), fysiotherapie (FT) en logopedie (LO) voor mensen met een neuromusculaire aandoening (NMA) gepresenteerd vanuit verschillende perspectieven: 1) professioneel perspectief; 2) wetenschappelijk perspectief; en 3) cliëntperspectief. Samen vormen deze perspectieven de elementen van evidence based practice en professioneel redeneren (*Hoofdstuk 1*).

De huidige paramedische zorg (ET, FT en LO) voor mensen met een NMA wordt gekenmerkt door grote variatie in verwijzing, kennis en ervaring bij verwijzers, bij professionals en bij cliënten. Dit proefschrift beoogt kennis en inzicht vanuit de eerdergenoemde perspectieven te vergroten.

Deel I van dit proefschrift representeert het **professionele perspectief**. In een cohort studie werd het gebruikelijke volume ET, FT en LO voor mensen met een NMA (referentiegroep n=106) vergeleken met het volume op basis van paramedisch advies vanuit een neuromusculair expertise centrum (interventiegroep n=102). Dit paramedisch advies was gebaseerd op eenmalige consulten ET, FT en LO en multidisciplinair overleg. In de referentiegroep werd bij aanvang het volume ET, FT en LO zowel retrospectief als prospectief vastgesteld met behulp van vragenlijsten. Daarnaast hielden patiënten gedurende acht weken in een zorgdagboek het volume ET, PT en LO bij. In de interventiegroep werden bij aanvang, na de consulten en na zes maanden vragenlijsten ingevuld door patiënten en professionals (ET, FT, LO en revalidatieartsen).

Bij de kostenanalyse (*Hoofdstuk 2*) werd gebruik gemaakt van de volumina en bijbehorende kosten van ET, FT en LO en de interventie (consulten en multidisciplinair overleg resulterend in een paramedisch advies). Bij vergelijking tussen de gebruikelijke zorg en geadviseerde zorg was sprake van een relatief ondergebruik van ET van 40% ten opzichte van de geadviseerde zorg. Bij FT was zowel overgebruik (32%) als ondergebruik (22%) en bij LO was geen ondergebruik, noch overgebruik. Bij 40% van de mensen werd tijdens het eenmalige LO consult advies gegeven zonder dat er een indicatie was voor verdere behandeling. Bij ET was dit bij 27% en bij FT bij 19% van de mensen. Wanneer de adviezen volledig werden opgevolgd zou de multidisciplinaire aanpak €85,- per persoon besparen (exclusief de kosten van de interventie €245,-). Het geadviseerde volume therapie was slechts gedeeltelijk geïmplementeerd na zes maanden. De conclusie is dat er enerzijds mensen met een NMA zijn die geen paramedische zorg krijgen, terwijl hier wel een indicatie voor is. Anderzijds zijn er mensen met een NMA

die continu therapie krijgen waarbij het de vraag is of deze behandeling verminderd dan wel volledig afgebouwd kan worden. Een aanbeveling is om de implementatie van de adviezen te bevorderen ten behoeve van een betere balans tussen kosten en baten.

Na zes maanden werd implementatie van de adviezen geëvalueerd voor ET, FT en LO. Ook werden factoren die mogelijk van invloed kunnen zijn nader geëxploreerd op niveau van het advies, de cliënt, de professional en de zorgorganisatie *(Hoofdstuk 3).* Het criterium voor succesvolle implementatie voor ET en LO was dat mensen ook daadwerkelijk therapie hadden gekregen wanneer dit was geadviseerd. Hierbij werd onderscheid gemaakt tussen adviezen voor ET en LO in de eerstelijn en in een revalidatiesetting. Voor PT werden andere criteria voor succesvolle implementatie toegepast, aangezien een aanzienlijk aantal mensen al FT had. Er werd onderscheid gemaakt tussen vijf categorieën: doorgaan *zonder* FT, doorgaan met huidige volume FT, stoppen met FT, volume FT verminderen, of starten met FT. Per categorie werd het percentage succesvolle implementatie berekend.

De resultaten lieten zien dat ET en LO adviezen volledig werden geïmplementeerd in de eerstelijn. De ET adviezen werden echter slechts gedeeltelijk (58%) geïmplementeerd in een revalidatiesetting. Bij FT werden de adviezen om met de huidige therapie door te gaan (n=64) volledig geïmplementeerd. Implementatie van de adviezen die een verandering van het therapievolume voorstelden was echter significant minder ($p < 0.001$). Van de adviezen om het volume te verminderen werd slechts 15% geïmplementeerd, van de adviezen om met de FT te stoppen 37.5% en van de adviezen om met FT te starten werd 29% geïmplementeerd. Mogelijke factoren die de implementatie hebben beïnvloed, waren gerelateerd aan het advies zelf (haalbaarheid van de therapieduur, correctheid en volledigheid), de persoon met NMD (motivatie) of de professional (ervaring in de behandeling van NMA). De therapeuten gaven de behoefte aan om het behandeladvies te bespreken met het multidisciplinaire team. Daarom werd aanbevolen voortaan contact op te nemen met de therapeuten om het advies te bespreken.

Deel II representeert het **wetenschappelijk perspectief** met drie systematische reviews naar respectievelijk de effectiviteit van ET, FT en LO bij volwassenen met NMA. Uitgebreide zoekstrategieën werden uitgevoerd voor zowel gecontroleerde als ongecontroleerde studies in de Cochrane Library, Medline, CINAHL en Embase databases. De zoekstrategieën bevatten

een zoekstrategie voor verschillende vormen van NMA, een zoekstrategie voor verschillende onderzoeksdesigns en zoekstrategieën voor ET *(Hoofdstuk 4)*, FT *(Hoofdstuk 5)* en LO *(Hoofdstuk 6)*.

Een best-evidence synthese werd toegepast waarbij drie niveaus van bewijsvoering werden onderscheiden: Niveau I bewijs verwijst naar tenminste twee RCTs (Randomized Controlled Trials) van voldoende methodologische kwaliteit. Niveau II bewijs verwijst naar één RCT van goede kwaliteit of tenminste twee onafhankelijke gecontroleerde studies (RCTs of controlled clinical trials , CCTs) van minder goede kwaliteit. Niveau III bewijs verwijst naar een RCT of CCT van minder goede methodologische kwaliteit of tenminste één studie met andere onderzoeksopzet van voldoende methodologische kwaliteit. Wanneer de studies van vergelijkbare opzet en methodologische kwaliteit leidden tot inconsistente bevindingen, werden de conlusies geformuleerd als "er is onvoldoende bewijs dat....".

Voor ET bij NMA werd niveau III bewijs gevonden dat training van de handfunctie bij spieraandoeningen effectief is. Er is geen bewijs gevonden voor de effectiviteit van ergotherapie interventies om de participatie van mensen met een NMA in betekenisvolle activiteiten en rollen te bevorderen. Voor PT is niveau II bewijs gevonden dat aerobe training in combinatie met krachttraining effectief is bij mensen met een spieraandoening. Er was niveau III bewijs dat aerobe training met of zonder krachttraining effectief is bij andere NMA. Ongecontroleerde studies resulteerden in niveau III bewijs voor de effectiviteit van ademhalingsoefeningen bij mensen met myasthenia gravis en myotone dystrofie. Voor LO bij NMA is slechts één ongecontroleerde studie gevonden op basis waarvan geconcludeerd wordt dat er niveau III bewijs is dat correctie van de hoofdhouding de efficiëntie van het slikken verbetert bij mensen met oculopharyngeale musculaire dystrofie (OPMD).

Deel III representeert **het cliëntperspectief.** Een kwalitatief onderzoek werd uitgevoerd met vijf echtparen waarvan drie mannen en drie vrouwen met de ziekte myotone dystrofie type 1 (MD1) en hun partners *(Hoofdstuk 7)*. Het doel was beter inzicht en begrip te krijgen hoe de ziekte MD1 het leven van echtparen beïnvloedt en hoe zij zich individueel en als echtpaar redden.

Alle echtparen werden tweemaal in de thuissituatie bezocht. Tijdens het eerste bezoek werden individuele diepte-interviews uitgevoerd en tijdens het tweede bezoek werd het echtpaar gezamenlijk geïnterviewd. De bevindingen zijn beschreven vanuit het perspectief van de persoon met MD1,

vanuit het perspectief van de partners en vanuit het perspectief van beiden als echtpaar.

De mensen met de ziekte ervaren het leven met de MD1 als het steeds opnieuw moeten inleveren van mogelijkheden. Hierbij beschrijven ze fysieke, cognitieve en psychosociale barrières bij de uitvoering van dagelijkse handelingen en sociale participatie. De partners benadrukten de toenemende belasting als mantelzorger. Verschillende partners gaven extra instructies, aanmoedigingen en geheugensteuntjes als strategie om te compenseren voor de verminderde initiatiefname en neiging om situaties te ontwijken. Als echtpaar werden ze geconfronteerd met de dilemma's en frustraties van het zoeken en aanpassen van een goede rolverdeling en het vinden van een balans tussen individuele en gezamenlijke activiteiten. Alle echtparen ervoeren onbegrip van familie, vrienden en de maatschappij met als gevolg dat mensen zich verder terugtrekken en in een isolement geraken. De gezondheidszorg werd ervaren als losse eilanden waarbij specialisten zich focussen op een stukje van de ziekte en niet begrijpen wat dit betekent voor het dagelijks leven van de mensen.

Deze studie laat zien dat mensen met MD1 en hun partners vele problemen ervaren waar in de gezondheidszorg onvoldoende aandacht aan wordt besteed. Vooral de impact van de cognitieve problemen en de sociale beperkingen wordt vaak onderschat. Echtparen ervaren achteruitgang als onvermijdelijk en als iets waar niets aan te doen is. De ervaren problemen worden vaak niet benoemd en er wordt vaak ook niet naar gevraagd. Communicatie met echtparen is dan ook essentieel om te achterhalen wat de impact van deze multisysteemziekte voor beiden is. Begeleiding van echtparen wordt aanbevolen waarbij beiden worden ondersteund in het omgaan met de gevolgen van de ziekte individueel en gezamenlijk als echtpaar.

In *Hoofdstuk 8* worden alle bevindingen bediscussieerd en wordt het Chronic Care Model (CCM) geïntroduceerd als bruikbaar model met elementen die de efficiëntie van paramedische zorg voor volwassenen met NMA kunnen verbeteren. Vooral verdere ontwikkeling en verbetering van de elementen zelfmanagement ondersteuning, optimaliseren van zorgprocessen, instrumenten voor besluitvorming en klinische informatiesystemen is wenselijk.

Conclusies en aanbevelingen

De huidige ET, FT en LO voor mensen met een NMA is niet efficiënt en paramedisch advies vanuit een neuromusculair centrum wordt gedeeltelijk geïmplementeerd. Daarnaast is de wetenschappelijke basis voor ET, PT en LO voor volwassenen met NMA beperkt. Tot slot blijken echtparen met MD1 veel meer beperkingen te ervaren in het dagelijks leven, dan wordt onderkend in de zorg. Daarom worden de volgende aanbevelingen gedaan voor de toekomstige paramedische zorg en onderzoek:

- Meer onderlinge uitwisseling van kennis en ervaring door professionals;
- Gebruik van gestandaardiseerde uitkomstmaten op het niveau van ziekten, lichaamsfuncties en structuren, activiteiten en participatie en persoonlijke en omgevingsfactoren;
- Ontwikkeling en implementatie van hulpmiddelen voor de klinische besluitvorming zoals richtlijnen voor verwijzing en voor ET, PT en LO voor mensen met NMA;
- Observationele studies met gebruik van klinische informatiesystemen waarin gegevens systematisch worden bijgehouden;
- Ontwikkeling, testen en implementatie van zelf-management programma's met als doel mensen met NMA en hun naasten te ondersteunen bij optimale participatie in betekenisvolle activiteiten en rollen; om dit te bereiken wordt het gebruik van het MRC raamwerk (vanuit de Medical Research Council) aanbevolen voor het ontwikkelen en testen van complexe interventies.

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logopediste en spraak-/taalpathologe Simone Knuijt. Gezamenlijk startten we het onderzoek getiteld 'gerichte verwijzing van mensen met een neuromusculaire aandoening naar paramedische zorg'. Dit onderzoek werd het SPADE onderzoek (Spierziekten Paramedisch Advies en Doelmatigheid in de Eerste lijn). Allan, we hadden de onderzoekstaken en aandachtsgebieden mooi verdeeld. Dankzij jou liep de logistiek gesmeerd en maakten we gebruik van de testorganiser voor alle vragenlijsten. Hartelijk dank! Simone, ook jij heel veel dank voor jouw grote aandeel bij alle studies evenals de logopedie review. Allan en Simone, geweldig dat jullie mijn paranymfen zijn.

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Curriculum Vitae

Edith Cup is op 6 mei 1966 geboren te Venray. Na de middelbare school (gymnasium ß scholengemeenschap Jerusalem te Venray) volgde ze van 1984 tot 1988 de opleiding ergotherapie in Hoensbroek. Na deze opleiding werkte ze als ergotherapeut in Engeland in verschillende instellingen in en rondom Coventry. In 1990 ging ze in het UMC St Radboud werken als ergotherapeut en in 1994 startte ze met een deeltijdstudie bewegingswetenschappen aan de VU in Amsterdam. Na de propedeuse maakte ze in 1995 de overstap naar de deeltijdstudie gezondheidswetenschappen, afstudeerrichting bewegingswetenschappen aan de Universiteit Maastricht. In 1998 studeerde ze cum laude af. Sindsdien combineert ze patiëntenzorg, onderzoek en onderwijs met als belangrijkste aandachtsgebied de ergotherapie bij volwassenen met chronisch neurologische aandoeningen, in het bijzonder CVA en neuromusculaire aandoeningen. Ze is auteur van meer dan 40 nationale en internationale publicaties op het gebied van de ergotherapie. Edith is getrouwd met Cees Kreulen en samen hebben ze twee dochters, Els (7) en Wies (5).

Stellingen

Behorend bij het proefschrift

Occupational therapy, physical therapy and speech therapy for persons with neuromuscular diseases

An evidence based orientation

1. *Meer* mensen met een neuromusculaire aandoening hebben veelal *minder* langdurig paramedische zorg nodig. (dit proefschrift)

2. Voor de implementatie van een paramedisch advies is meer nodig dan alleen het versturen ervan. (dit proefschrift)

3. In tegenstelling tot eerdere opvattingen is matig intensieve spierkrachttraining bij de meeste spierziekten niet schadelijk, maar veel sterker word je er niet van. (dit proefschrift)

4. De 'kracht' die doorgaans wordt gemeten, zegt niets over de 'kracht' die mensen hebben om met hun beperkingen om te gaan.

5. Om de effectiviteit te bepalen van paramedische zorg bij mensen met een neuromusculaire aandoening kan niet alleen worden volstaan met een RCT. (dit proefschrift)

6. Onbegrip en miscommunicatie zijn niet altijd het gevolg van dysartrie; vaak is het een kwestie van goed luisteren. (dit proefschrift)

7. Bij myotone dystrofie wordt de impact van de psychosociale problemen voor mensen met de ziekte, hun partners en voor beiden als echtpaar vaak zwaar onderschat. (dit proefschrift)

8. Niet alles wat geteld kan worden telt, en niet alles wat telt kan geteld worden. (Albert Einstein)

9. Fietsen door Heumensoord (omslag) helpt om door het bos de bomen te zien.

10. The quality of a life can best be appraised by the person whose life it is. (Karen Whalley Hammel)

11. Met geluk is het net als met gezondheid; als je er niets van merkt, betekent dit dat het er is. (Iwan Toergenjew; ter nagedachtenis aan Anjorieke Richt)