

Physiotherapy in Huntington's disease: Guidance for Practice

Fizjoterapia w chorobie Huntingtona: Wytyczne dla praktyki

CONTRIBUTING MEMBERS OF THE EUROPEAN HUNTINGTON'S DISEASE NETWORK (EHDN) PHYSIOTHERAPY WORKING GROUP

GUIDANCE DOCUMENT: LORI QUINN^{1/}, MONICA BUSSE^{1/}, ANNE-WIL HEEMSKERK^{2/}, CAMILLA EK WALL^{3/}, NORA FRITZ^{4/}, DEBORAH KEGELMEYER^{4/}, ANNE KLOOS^{4/}, ASHWINI RAO^{5/}, MAGGIE BROAD^{6/}, HELEN DAWES^{7/}, CAROL HOPKINS^{8/}, UNA JONES^{1/}, HANAN KHALIL^{1/}, CHARMAINE MEEK^{9/}, JANE OWEN^{10/}, RUTH SANDS^{11/}, SHEILA WATTERS^{12/}, CATHERINE SACKLEY^{9/}, PAOLA ZINZI^{13/}

TREATMENT BASED CLASSIFICATIONS: LORI QUINN^{1/}, MONICA BUSSE^{1/}, HANAN KHALIL^{1/}, UNA JONES^{1/}, ANGELA HALL^{14/}, SUE ARMSTRONG^{14/}, KARIN BUNNIG^{2/}, MAGGIE BROAD^{6/}, KATY DEBONO^{1/}, CAMILLA EK WALL^{3/}, HANNE FOSSMO^{15/}, NORA FRITZ^{4/}, KAREN JONES^{1/}, UNA JONES^{1/}, DEB KEGELMEYER^{4/}, HANAN KHALIL^{1/}, ANNE KLOOS^{4/}, RODOLFO VERA^{13/}, ALEXANDRA SCHULER^{16/}, JESSIE VAN DER BENT^{2/}, AND RODOLFO MARCELO VERA URRESTARAZU^{17/}

Affiliations of contributing members:

- ^{1/} School of Healthcare Studies, Cardiff University, Cardiff, United Kingdom
- ^{2/} Huntington Centre Overduin, Katwijk, Nederland
- ^{3/} University Hospital, Department of Physiotherapy, Uppsala, Sweden
- ^{4/} The Ohio State University, College of Medicine, Physical Therapy Division, Columbus, USA
- ^{5/} Columbia University Medical Center, Department of Rehabilitation Medicine, Neurological Institute, New York, USA
- ^{6/} Community Neurological Team, Worthing, West Sussex, United Kingdom
- ^{7/} Oxford Brookes University, Movement Science Group, Oxford, United Kingdom
- ^{8/} The Royal Hospital for Neuro-disability, Physiotherapy Department, London, United Kingdom
- ^{9/} Primary Care Clinical Sciences, School of Health and Population Sciences, College of Medical and Dental Sciences University of Birmingham
- ^{10/} Plympton Hospital, Therapies Office, Plympton, Plymouth, Devon, United Kingdom
- ^{11/} Regional Care Advisor – North and South East London, HDA, Neurosupport Centre, Liverpool, United Kingdom
- ^{12/} Bristol General Hospital, Disabled Adult Resource Team, Bristol, United Kingdom
- ^{13/} CNR – Consiglio Nazionale delle Ricerche, Italy
- ^{14/} Community Huntington's Disease Team, Aneurin Bevan Health Board, United Kingdom
- ^{15/} Vikersund Kurbad AS
- ^{16/} Gümligen, Bern, Switzerland
- ^{17/} Asociación Corea de Huntington Española, Josep Barges I Barba, Calafell, Tarragona, Spain

Wprowadzenie. Choroba Huntingtona (HD) jest schorzeniem neurodegeneracyjnym charakteryzującym się triadą objawów obejmującą zaburzenia ruchowe, zachowania i poznawcze. Brak jest obecnie leczenia modyfikującego przebieg HD, ale niektóre badania nie spełniające kryteriów EBM, sugerują, iż fizjoterapia może przynieść pacjentom korzyści w zakresie codziennego funkcjonowania oraz poprawę jakości życia. Grupa Robocza Fizjoterapii (PWG), Europejskiej Sieci Choroby Huntingtona (EHDN) postanowiła, zatem stworzyć oparty na dowodach przewodnik, aby wesprzeć zastosowanie programu fizjoterapii strukturalnej w ujednoczony, oraz dostosowany do potrzeb pacjentów sposób w zależności od stanu zaawansowania HD.

Metody. W sposób planowy dokonano przeglądu literatury. Z uwagi na niedobór pozycji piśmienniczych umożliwiających pełną ocenę metod fizjoterapeutycznych pod kątem zastosowania fizjoterapii w HD, spełniającą kryteria EBM, zostały uformowane podgrupy ekspertów, których zadaniem było wypracowanie konsensusu dotyczącego najlepszej praktyki klinicznej. Szkic stworzonego dokumentu został przesłany pełnoprawnym członkom PWG, innym fizjoterapeutom i innym profesjonalistom w ochronie zdrowia z EHDN, aby pozyskać informacje zwrotną i komentarze.

Wyniki. Utworzono przewodnik dotyczący 8 sfer postępowania fizjoterapeutycznego w HD. Aby ułatwić wykorzystanie praktyczne powstałego dokumentu klinicytom, został utworzony system kwalifikacji pacjentów na podstawie ich objawów służący dostosowaniu odpowiedniej metody leczenia. Prowadzi to do utworzenia i rozwoju bardziej ujednoczonego podejścia do terapii.

Dyskusja. Przewodnik fizjoterapii w HD jest zbiorczym, opartym na konsensusie i dowodach dokumentem, który może być wykorzystany przez fizjoterapeutów, aby wprowadzić program opieki dla pacjentów w zależności od klinicznego zaawansowania HD, ułożony według zasad najlepszej praktyki klinicznej. Gdy tylko dostępne staną się nowe dowody będzie konieczne przeprowadzenie ponownego ich przeglądu celem modyfikacji przewodnika. System dostosowania odpowiedniej metody leczenia, który ma na celu lepszą kategoryzację objawów oraz powiązanie ich z właściwym planem terapeutycznym może być użyteczny, aby dokonać klinicznej argumentacji i efektywnej oceny wyników w leczeniu stosunkowo rzadkich schorzeń takich jak HD.

Słowa kluczowe: choroba Huntingtona, fizjoterapia, terapia fizykalna, interwencja, przewodnik kliniczny, system dostosowania odpowiedniej metody leczenia

Background. Huntington's Disease (HD) is a degenerative neurological disease that results in a triad of clinical symptoms, encompassing motor, behavioural and cognitive impairments. To date there are no disease-modifying treatments for HD, but some uncontrolled studies suggest that physiotherapy may produce functional benefit with resulting improvement daily functioning and quality of life. The Physiotherapy Working Group (PWG) of the European Huntington's Disease Network (EHDN) therefore set out to develop evidenced based guidance to support uniform implementation of a structured physiotherapy programme according to clinical need across the stages of the condition.

Methods. A review of the literature was conducted using a systematic approach. There was insufficient literature in support of physiotherapy interventions and approaches to be able to conduct a complete evidenced based review, therefore physiotherapy expert sub-groups were formed to incorporate consensus as to best practice. A draft document was distributed to the entire membership of the working group, to outside physiotherapists and other health care professionals within EHDN to elicit feedback and comments.

Results. A Guidance Document covering 8 specific areas pertaining to physiotherapy management of HD was developed. In order to facilitate the document's practical usability amongst clinicians, a treatment-based classification system was further proposed to allow categorization based on presenting signs and symptoms. This would provide a foundation for development of a more uniform approach to intervention delivery.

Discussion. The Physiotherapy Guidance for HD is a comprehensive, consensus- and evidence-based document that can be used by physiotherapists to implement a plan of care that is currently consistent with best practice for individuals at all stages of HD. As evidence becomes available, future systematic reviews will be required in order to inform further development. The use of treatment-based classifications, which aim to better categorize common signs and symptoms and link them to appropriate intervention plans, may be useful in relatively rare diseases such as HD to aid clinical reasoning and promote effective outcome evaluation.

Key words: Huntington's disease, physiotherapy, physical therapy, intervention, clinical guidelines, treatment based classifications

Introduction

Huntington's disease (HD) is a dominantly inherited progressive neurodegenerative condition caused by a CAG repeat mutation in the HTT gene. It predominantly affects the brain, causing dysfunction and death of medium spiny striatal projection neurons and thus disruption of corticostriatal pathways with resultant impairment of cognition, motor function, and behaviour. To date there are no disease-modifying treatments for HD, but some uncontrolled studies suggest that physiotherapy may produce functional benefit with resulting improvement daily functioning and quality of life. Provision of physiotherapy intervention is however not consistent [1]. The beneficial role of physiotherapy for people with Parkinson's Disease, a basal ganglia disorder with some similar features to HD, has been well illustrated,

with two recent systematic reviews reporting that physiotherapy can improve multiple factors including physical functioning, health-related quality of life (HR-QoL), strength, balance and gait [2, 3].

The literature in support of physiotherapy for people with HD is less clear. Two reviews have noted that to date there has been only a small amount of evidence in support of physiotherapy within HD [4, 5]. Although the impact of currently available literature is somewhat overshadowed by poor methodological rigour, small sample sizes, unclear selection criteria (resulting in potential heterogeneity in participant groups), and a lack of follow-up, there have more recently been some optimistic developments. For example, a before-after trial with a sample size of 40 found an intensive rehabilitation programme of 6 sessions per week demonstrated an improvement

in motor function over the two year period [6, 7]. Positive findings from environmental enrichment studies in mice also provide some support for the basis of physiotherapy for people with HD. In mice models of HD, those mice placed within an environment providing physical, mental and social stimulation, have a slower disease progression, and maintain motor function for longer compared to those housed in more isolated conditions [8, 9]. People with HD have a range of physical, cognitive, psychological and social care needs over an extended time frame. One of the difficulties in developing clinical guidelines for complex neurodegenerative diseases such as HD is the heterogeneity of clinical signs and symptoms. Whilst staging of the disease process (e.g. early, middle, late) can provide a general framework for intervention, within each stage there is a wide range of potential impairments that can impact on an individuals' level of functional activity and life participation. This makes structuring of consistent therapeutic approaches problematic. This problem is not unique to HD, and has been documented in other physiotherapy patient groups, most notably low back and neck dysfunction, and for general neurorehabilitation patient groups [10-15]. Creating sub classifications of patients' impairments and problems, and matching those with more specific physiotherapy interventions may improve goal setting and consequently clinical outcomes [10-12, 15]. Clinical guidelines are evidenced-based recommendations for clinical practice in specific conditions [16, 17]. The availability of clinical guidelines facilitates uniformity of care and standards of practice with the aim of improving quality of care provision. Use of appropriate guidelines and sub-classifications can change the process and practice of care for the benefit of the patient and can inform research process leading to more effective research into practice. It is critically important for health professionals to be able to define and document their intervention strategies and evaluation practices, in line with clinical guidelines so that systematic outcome evaluation can be done and best practice highlighted.

Clinical guidelines are in place for physiotherapy in other neurodegenerative diseases, such as Parkinson's Disease [13, 18] and Multiple Sclerosis [19]. Whilst physiotherapy is increasingly being proposed as a mode of intervention for people with HD, to date there has been no formalized clinical guidelines or standards of care to guide practice. The Physiotherapy Working Group (PWG) of the European Huntington's Disease Network therefore set out to develop a guidance document, to provide a written framework for the physiotherapeutic management of people with HD. The aims were to provide, where possible, a scientific

evidence-based document to inform the optimal, individualised physiotherapeutic management of people with HD. This included the provision of a written framework for the physiotherapeutic management of people with HD, in order to enable uniformity of care internationally. We also aimed to highlight the scientific evidence for PT practice with people with HD, evaluating the effectiveness of any intervention strategies presented in the literature. A guidance document would allow practitioners to make an informed decision regarding patient care and to highlight areas within practice that currently lack the scientific evidence base to inform future research.

Methods

The Physiotherapy Guidance Document for HD was created through a combination of available scientific evidence and expert consensus. A PICO (Population, Intervention, Comparison, and Outcome) approach to searching the literature was used. The population was defined as people with Huntington's Disease. The intervention was any physiotherapy-based intervention or assessment. No set comparisons were made or follow up times set. Outcomes were any reliable and valid measure of physiotherapy – related interventions.

Databases searched were Ovid MEDLINE(R) 1950 to March Week 2 2009; EMBASE 1980 to 2009; Week 12, 21st March 2009; EBSCO CINAHL 1981 to 21st March 2009; PEDro 1929 to 21st March 2009. Search terms included: Huntington's disease, Chorea, Huntington, Huntington Chorea; Physical therapy modalities, physical therapy, Physiotherapy, Rehabilitation / rehabilitation, occupational therapy / occupational therapy, Physical activity, exercise therapy/ exercise therapy, Activities of daily living/ activities of daily living, physical mobility, muscle stretching exercises/ stretching, respiratory therapy/ respiratory therapy, flexibility, range of motion, range of movement, upper extremity/ upper limb, reaching, reach, grasping, grasp, reach to grasp, posture/ posture, balance, accidental falls,/ falls, equipment and supplies/ equipment, seating, positioning, transfers, functional status/ functional status, gait/ gait.

Inclusion criteria were: articles published in English; therapy in human HD subjects only; subjects to be of 18 years of age + with a confirmed diagnosis of HD; study specific to physiotherapy/occupational therapy/rehabilitation and exercise. A total of 239 individual papers were identified through data base searching. Two hundred and six of these were excluded leaving a total of 33 papers for review. The main reasons for exclusion were for non-human studies, subjects did not have HD, and the study was not related to physiotherapy/occupational therapy/rehabilitation or

exercise. Two of these were not retrieved and 31 were summarised in the full guidance document. Three reviews, 9 PT related studies, 11 gait specific, 1 muscle strength, 3 balance and mobility, 1 dystonia, 2 upper limb specific and 1 paper regarding multi-sensory stimulation were identified.

The identified evidence was then summarised independently and then discussed at a European Huntington's Disease Network (EHDN) Physiotherapy Working Group (PWG) meeting. This summary is presented in the final guidance document. Due to the paucity of scientific evidence, recommendations were also formulated based on expert consensus from the EHDNPWG. Following completion of the initial draft document, the Guidance Document was disseminated to all members of the PWG and other interested healthcare professionals. Amendments were made to the document based on their feedback. In a second phase of development, sections of the Guidance Document were reviewed in detail by subgroups of 2-3 members of the PWG. Feedback from the subgroups were directed back to the authors, and integrated into the final document.

Following publication and dissemination of the Guidance Document, the PWG met and discussed the impact of the document and provided constructive feedback. A consensus was reached that the next step in development of Guidance for Physiotherapists was to provide more specific guidelines within a user-friendly format. The requirements for a subsequent document were that it had a simple, organized structure that made it readily useful to novice therapists, but one that would not be limiting or overly prescriptive for more experienced clinicians. The methodology provided by treatment-based classification systems was an obvious choice for structuring such guidance. The group agreed that current research in the area of treatment-based classification [10-12, 15] had potential for application to the HD population. The group proposed a range of treatment-based classifications to address the heterogeneity of patient conditions, to provide structure for standardised data collection of interventions and outcomes, and consequently inform evaluation of complex interventions and advance research into care and evidenced-based service delivery for people with HD. A second sub-group was subsequently formed, consisting of expert physiotherapists within HD and neurodegenerative diseases. Two focus group meetings of this group were held over a 6 week period, during which time treatment based classifications were constructed using the Guidance Document as a template for discussions. An iterative process was undertaken where key impairments were classified, so that the key signs and symptoms commonly found

in HD could be organized within a classification system. The discussions also focussed on linking these classifications with specific interventions and outcome measures from the Guidance Document.

Results: Physiotherapy Guidance for HD

The review of the literature revealed a lack of any well controlled randomised intervention studies of physiotherapy interventions in HD that could be used to conduct a systematic review. We considered all relevant literature and case reports as well as observational studies that were identified in the review to provide some insight as to the nature of the impairments that could provide a theoretical foundation and recommendations for physiotherapy interventions. Alongside this process of reviewing the literature, we consulted with expert clinicians to finalise our recommendations.

A brief overview of the philosophy and recommendations for developing physiotherapy plans as well as considerations of procedural interventions is presented here under the following sub-headings: Framework, Evaluation and Outcome Measures, Interventions, Documentation and Interdisciplinary management, Cost Implications, Treatment-based classifications and Dissemination. Here we report the summary recommendations. Greater detail on each topic as well as the review of the literature can be found in the formal physiotherapy guidance document: <https://www.eurohd.net/html/network/groups/physio/physiotherapy-guidance-doc-2009.pdf?eurohdsid=d7e987cc87f9d541b70dc6497d223d66>

Framework for patient management. An important component to patient management in a neurodegenerative disease such as HD is to consider people at all stages of the condition, including those who have the mutation for HD but are not displaying motor symptoms (pre-manifest) [1, 20]. It is further suggested that physiotherapy management of people with HD should be modified according to individual problems and to the stage of the disease. The reader is referred to the original paper where the framework development is described [1]. Currently, intervention focuses on symptomatic management, however there is increasing support for early intervention where an impact may be made on biological processes with the potential to influence on the natural history of the condition. In addition, early referral to PT for people with HD may be beneficial in a number of ways. It enables practitioners to ascertain a baseline for the person with HD, supports the establishment of a therapeutic relationship between the person with HD, practitioners and caregivers, and ensures early intervention to attempt to maintain mobility and function for as long as possible [1].

Evaluation and Outcome Measures. The physiotherapy evaluation for people with HD should be both anticipatory and responsive to the disease stage. The physiotherapist should work in collaboration with the patient and evaluate their individual considerations [21]. The World Health Organisation International Classification of Functioning, Disability and Health (ICF) [22] can aid assessment of a person's level of functioning, and further facilitate consideration of the triad of motor, cognitive and psychiatric symptoms that are often seen in HD. It can be used also for goal setting & treatment planning, and focuses on aspects of a person's health and health-related wellbeing in terms of activities and participation i.e. the description of the tasks (activities) or life situations (participation) the person wishes to be involved in, and the impact that impaired body function or structure is having on these aspects.

A structured evaluation for the patient with HD is comprised of both subjective and objective assessments, the primary goal being to determine the relationship between any impairments, activity limitations and participation restrictions. Caregiver involvement, wherever possible, is strongly encouraged, particularly if the patient has difficulties communicating through cognitive or physical impairment. The objective evaluation should include assessment of neuromuscular (e.g. bradykinesia, dystonia, chorea), musculoskeletal (e.g. posture, range of motion, pain, muscle strength), and cardio-respiratory and cardio-vascular impairments (e.g. vital signs, breathing function, exercise capacity). Standardized tests can be utilized as outcome measures that may be sensitive to changes related to any physiotherapy intervention. The Berg Balance Scale, Tinetti Mobility Scale, Timed Up and Go Test, Four square step test and the Functional Reach Test have all been found to be valid and responsive clinical measures in HD and are considered useful to detect those at risk of falls [23-26].

Physiotherapy Interventions. Since pharmacologic interventions to date have been ineffective in altering or slowing the disease process, healthcare practitioners and patients themselves continue to look for alternative therapies. While physiotherapy has been shown to be of benefit for some neurodegenerative diseases, such as PD and MS, the evidence supporting its use for people with HD is limited. Nonetheless, utilizing traditional physiotherapy techniques with applicability to the special needs of this population may provide some benefit for a disease that currently lacks any effective treatment regimes.

The main goals of physiotherapy intervention will generally change over time. In the early stages,

interventions will be primarily preventative. As the disease progresses, interventions become restorative in nature, addressing specific physical impairments and resulting activity limitations. In the later stages, intervention is primarily compensatory, in which therapists develop strategies to maximize a patient's functional ability despite progression of the disease. With regards to falls risk, a clear justification of benefit of any particular management strategy compared to the risk that the situation presents should be elucidated. Therapists working with people with HD should acknowledge and facilitate independent mobility for as long as is reasonably possible [1].

Goals should be specific and measurable and mutually agreeable to both the patient and the therapist [27]. They should address specific functional problems or participation restrictions that are amenable to physiotherapy intervention. Goals to decrease chorea or dystonia, for example, may not be realistic. Furthermore, amelioration of any particular impairment in HD may not translate into functional improvements, and therefore therapists should focus on functional gains while attempting to ascertain the influence various impairments have on activity limitations in each of their patients.

Similar to healthy individuals, exercise, including strengthening and cardiovascular conditioning, is recommended for people who are pre-manifest and in the early stages of the disease. Exercise may be helpful in minimizing any neuromuscular or musculoskeletal effects that come with disease progression. As the disease progresses and specific problems become evident, therapists must begin to ascertain those impairments that may be contributing to specific activity limitations or participation restrictions. For example, involuntary movements could contribute to balance problems, but inactivity may also be a contributing factor. Impairment-based diagnoses that may impact on function include: dystonia; bradykinesia; moderate-severe chorea or ballismus; rigidity; impaired respiratory function and fatigue. It is important to manage the secondary effects of such impairments. For example, for patients with chorea, protective equipment can be provided, and for patients with dystonia, loss of range of motion and muscle imbalance should be prevented. Functional problems that may occur include: impaired fine motor skills and manual dexterity, impaired sitting posture and sitting ability, impaired mobility, transfers and gait; impaired balance/ and risk of falls and reduced cardiovascular and general physical fitness [1, 23, 24, 25]. In general, it is thought that physical fitness plays a part in (i) a patient's motivation to keep as active as possible and (ii) their ability to cope with more challenging treatment interventions.

The potential impact of psychiatric impairments and a patient's cognitive status cannot be overlooked. Therapists should consider whether or not a patient has memory loss, depression, aggression, obsessive-compulsive tendencies, or anxiety, to name a few, and these impairments must be taken into account during the design of any intervention plan [28].

Documentation and interdisciplinary management. Developing and documenting a plan of care for the patient with HD should also include coordination of services and communication that needs to take place with other professionals, caregivers or family members so that the physiotherapy plan can be implemented. This may also include referrals to other professionals and additional documentation of any planned home programme and patient/caregiver education. Importantly, the physiotherapist should be part of an interdisciplinary team that is involved in the management of patients with HD throughout the lifespan of their disease. Such professionals include geneticists, neurologists, nurses, occupational therapists, psychologists, speech language therapists, and social workers, among others. It is critical for therapists to consider collaboration with these professionals and to understand their role as part of an interdisciplinary team and in particular their contribution to palliative care in the late stages of the disease. The ultimate aim for palliative care is to achieve the best possible quality of life for patients and their families [29]. The physiotherapist can advise on seating and positioning, respiratory management as well as relaxation techniques and may also have a role to play when considering advanced care directives [30].

Cost implications. It is also important for physiotherapists to consider the cost implications of their services throughout a life-long disease. It is unreasonable to expect that any funding agency (government or otherwise) would be agreeable to ongoing physiotherapy intervention 2-3 times/week for the extended disease process. Instead, therapists should consider serving consultative roles to patients from an early stage, and providing more intensive intervention when a change in functional status warrants it.

Treatment-based classifications. Seven treatment-based classifications specific to HD have been developed using the Guidance document as a reference point. The primary findings discussed above for the Guidance document were utilized to categorize patients more specifically based on primary impairments and activity limitations (see Table I). Within each classification, we determined signs and symptoms as a method for categorization. We then listed general aims, and provided specific intervention

strategies and outcome measures. Examples of classifications are available from the authors, and will be made available in full on the EHDN website in due course. Clinical validation of these classifications is now required.

Table I. Seven treatment-based classifications developed for physiotherapy management of people with HD

Classification	Description of clinical findings	Stages of Disease
A. Exercise Capacity and Performance	Absence of motor impairment; potential for cognitive and/or behavioural issues	Presymptomatic/early
B. Planning and sequencing of tasks (including bradykinesia)	Difficulty and slowness in performing functional activities (dressing, bathing, ADLs, sit to stand, etc). Presence of apraxia or impaired motor planning; slowness of movement and/or altered force generation capacity	Early-mid
C. Mobility, Balance and Falls Risk	Ambulatory for community and/or household distances; balance, strength or fatigue resulting in falls or high risk for falls	Early-mid
D. Secondary adaptive changes and deconditioning	Musculoskeletal and/or respiratory changes resulting in decreased participation in daily activities	Early-mid
E. Impaired Postural Control and alignment in sitting	Inappropriate alignment due to adaptive changes, involuntary movement, inability to facilitate or coordinate movement	Mid-late
F. Respiratory dysfunction	Impaired respiratory function and capacity; limited endurance; impaired airway clearance; risk for infection	Mid-late
G. Palliative Care	Unable to ambulate; dependent for most ADLs; difficulty maintaining upright sitting position; range of motion and pulmonary issues	Late

Dissemination. In order to implement and achieve standardization of practice, the PWG decided that it was critical for this work to be made freely available and shared as widely as possible. This was important also to elicit feedback from physiotherapists and other healthcare professionals working in the field. We therefore published 2000 print copies of the Guidance Document, which were distributed to all EHDN sites, physiotherapy education programmes in the UK, HD Associations across Europe, and to other clinical and academic sites upon request. This work has also been presented at numerous conferences as well as family and healthcare professional information days across the UK and Europe. The treatment based classifications are in the process of being translated by EHDN language co-ordinators across the network and will be available for download from the physiotherapy working group web pages in due course.

Discussion

While physiotherapy is increasingly being considered an important mode of intervention for people with HD, to date there has been no formalized guidance or standards of care to document best practice. A need was identified from within the HD community to first and foremost conduct pragmatic analysis of current physiotherapy service delivery in HD, and to document the available evidence related to physiotherapy services. This paper describes the process and outcomes from the development of these clinical guidelines for physiotherapy management of people with HD.

Due to the progressive nature of HD, the needs of people with this condition change over time, and the role of the physiotherapist should be flexible in response to the clinical need over the prolonged course of the illness. Although the evidence supporting physiotherapy interventions for people with HD is limited, there is some suggestion from HD literature and from other similar neurodegenerative diseases that exercise and other physiotherapy strategies may be useful in minimizing the effects of this devastating disease. The Physiotherapy Guidance Document has been developed based on expert consensus and available evidence and is freely available to the HD community. It does, however, continue to be a work in progress and will require review and updating as new more robust research becomes available.

The availability of a guidance document is the first step towards standardization of physiotherapy practice. We must however highlight that the background research done to support the guidance document yielded a paucity of applicable research studies, and no randomized controlled trials aimed at evaluating the efficacy of physiotherapy or exercise. The challenge of evaluating physiotherapy is largely due to problems of identifying, documenting and reproducing the multiple components of the intervention. The randomised controlled trial was originally developed for evaluation of single interventions such as a particular drug, and is not fully compatible with complex interventions such as physiotherapy. It is clear that rigorous evaluation of complex interventions such as physiotherapy is imperative to their continued existence, but this requires a phased approach that can ultimately lead to a definitive controlled trial [31]. The Medical Research Council Framework for evaluation of complex interventions [31] refers to a theoretical and a modelling stage prior to conducting full scale randomised controlled clinical studies. The first step is to review the relevant theory and literature that is available to ensure best design of intervention; this is

called the theoretical phase. The next modelling phase involves identifying components of the intervention of choice (based on the previous theoretical stage) and determining how the outcomes would be achieved. It is clear that mixed methodology incorporating both quantitative and qualitative methods are relevant to these initial stages. The Guidance Document developed for HD used this phased approach and provides a starting point for the theoretical and modelling components of the framework. It provides the foundations for development of future physiotherapy related controlled trials.

Development of treatment-based classifications as an extension of the Guidance Document can help to further facilitate standardization of practice. This approach requires an expansion and some modification of the conceptual framework presented [1] which bases intervention strategies on stages of the disease [1, 20]. Treatment-based classifications address the issue of heterogeneity within HD by targeting specific problems that are seen irrespective of stage of disease. This allows for a more individualized approach to patient management but incorporates a clear structure for evaluation of efficacy.

Limitations, Implications and Future Directions

The development of this Guidance Document and the subsequent classification system is the first step in the process towards solidifying the evidence base in physiotherapy in HD. The literature review for this document was limited to English speaking articles, and thus should be a starting point for a more extensive multi-language search. An important next step is clinical validation and revision of the classifications, in an effort to promote standardization of care. Once standardization of care is achieved, we can take the next important step in evaluating the efficacy of our interventions through pragmatic controlled clinical trials.

Acknowledgements:

The authors would like to acknowledge the contributing members of the European Huntington's Disease Network (EHDN) Physiotherapy Working Group to the writing of the guidance document. The authors would also like to acknowledge Raymond Roos and Sheila Simpson for their input and the EHDN for their support in funding physiotherapy working group meetings.

Financial Disclosure: The EHDN had no role in study design, data collection and analysis, decision to publish, or preparation of the manuscript.

Piśmiennictwo / References

1. Busse M, Khalil H, Quinn L, Rosser A. Physical therapy intervention for people with Huntington Disease. *Phys Ther* 2008, 88(7): 820-831.
2. Goodwin V, Richards SH, Taylor RS, Taylor AH, Campbell JL. The effectiveness of exercise interventions for people with Parkinson's disease: A systematic review and meta-analysis. *Mov Disord* 2008, 23(5): 631-40.
3. Kwakkel G, de Geode CJT, van Wegen EEH. Impact of physical therapy for Parkinson's disease: A critical review of the literature. *Parkinsonism and related disorders*. 2007, 13: S478-S87.
4. Bilney B, Morris M, Perry A. Effectiveness of physiotherapy, occupational therapy, and speech pathology for people with Huntington's disease: a systematic review. *Neurorehab Neural Repair* 2003, 17(1): 12-24.
5. Bilney B, Morris M, Denisenko S. Physiotherapy for people with movement disorders arising from basal ganglia dysfunction. *New Zealand J Physiotherapy* 2003, 31(2): 94-100.
6. Zinzi P, Salmaso D, De Grandis R, et al. Effects of an intensive rehabilitation programme on patients with Huntington's disease: a pilot study. *Clin Rehabil* 2007, 21(7): 603-613.
7. Zinzi P, Salmaso D, Frontali M, Jacopini G. Patient's and caregivers' perspectives assessing an intensive rehabilitation programme and outcomes in Huntington's disease. *J Publ Health* 2009, 17(5): 331-8.
8. Dobrossy M, Dunnett S. Optimising plasticity: environmental and training associated factors in transplant-mediated brain repair. *Rev Neurosci* 2005, 16(1): 1-21.
9. Hockly E, Cordery PM, Woodman B, et al. Environmental enrichment slows disease progression in R6/2 Huntington's disease mice. *Annals Neurol* 2002, 51(2): 235-242.
10. Scheets PL, Sarhmann SA, Norton BJ. Use of movement system diagnoses in the management of patients with neuromuscular conditions: a multiple-patient case report. *Phys Ther* 2007, 87: 654-669.
11. Delitto A, Erhard RE, Bowling RW. A treatment-based classification approach to low back syndrome: identifying and staging patients for conservative treatment. *Phys Ther* 1995, 75: 470-485.
12. Fritz J, Clelan JA, Childs J. Subgrouping Patients With Low Back Pain: Evolution of a Classification Approach to Physical Therapy. *J Orthop Sports Phys Ther* 2007, 37(6): 290-302.
13. Deane K. Physiotherapy for Parkinson's disease: a comparison of techniques. *Cochrane Database of Systematic Reviews*. 2001, 1: 1-20.
14. Pomeroy V, Niven DS, Barrow S, Faragher EB. Unpacking the black box of nursing and therapy practice for post-stroke shoulder pain. *Clin Rehab* 2001, 15(1): 67-83.
15. Fritz JM, Brennan GP. Preliminary examination of a proposed treatment-based classification system for patients receiving physical therapy interventions for neck pain. *Phys Ther* 2007, 87: 513-524.
16. Shekelle PG, Woolf SH, Eccles M, Grimshaw J. Clinical guidelines: developing guidelines. *BMJ* 1999, 318: 593-596.
17. Woolf S, Grol R, Hutchinson A, Eccles M, Grimshaw J. Clinical guidelines: Potential benefits, limitations, and harms of clinical guidelines. *BMJ* 1999, 318: 527-530.
18. Keus S, et al. Clinical practice guidelines for physical therapy in patients with Parkinson's disease. *Dutch J Physiotherapy* 2004, 114(3 suppl): 1-94.
19. Freeman J. Scoping Exercise for the MS Society. *Evolving Health Services Research Programme. Physiotherapy in Multiple Sclerosis*. MS Society, London 2005.
20. Dal Bello-Haas V. A framework for rehabilitation of neurodegenerative disease: planning care and maximizing quality of life. *Neurol Rep* 2002, 26(3): 115-129.
21. Quinn L, Busse M, Khalil H, et al. Client and therapist views on exercise programmes for early-mid stage Parkinson's disease and Huntington's disease. *Disabil Rehabil* 2010, 32(11): 917-28.
22. World Health Organization International Classification of Functioning, Disability and Health: World Health Organization, Geneva 2001.
23. Rao A, Muratori L, Louis ED, Moskowitz CB, Marder KS. Clinical measurement of mobility and balance impairments in Huntington's disease: validity and responsiveness. *Gait Posture* 2009, 29(3): 433-6.
24. Grimbergen YA. Falls and gait disturbances in Huntington's disease. *Mov Disord* 2008, 23(7): 970-6.
25. Busse ME, Wiles CM, Rosser AE. Mobility and falls in people with Huntington's disease. *J Neurol Neurosurg Psychiatry* 2009, 80(1): 88-90.
26. Quinn L, Rao A. Physical therapy for people with Huntington disease: current perspectives and case report. *Neurol Rep* 2002, 26(3): 145-53.
27. Randall KE, McEwen IR. Writing patient-centered functional goals. *Phys Ther* 2000, 80(12): 1197-203.
28. Imbriglio S. Huntington's disease at mid-stage. *Clinical Management* 1992, 12(5): 62-72.
29. Moskowitz CB, Marder K. Palliative care for people with late-stage Huntington's disease. *Neurol Clin* 2001, 19(4): 849-865.
30. Simpson SA. Late stage care in Huntington's disease. *Brain Research Bulletin* 2007, 72(2-3): 179-181.
31. Campbell M, Fitzpatrick R, Haines A, et al. Framework for design and evaluation of complex interventions to improve health. *BMJ* 2000, 321: 694-696.
32. Orth M, Handley OJ, Schwenke C, et al. Observing Huntington's Disease: the European Huntington's Disease Network's REGISTRY. *PLoS Curr* 2, pii: RRN1184, 2010.