Rehabilitation of Huntington's disease

Citation:

This version is free to view and download for private research and study only. Not for re-distribution or re-use.

©2013, Cambridge University Press

Reproduced with permission.

Downloaded from DRO:
http://hdl.handle.net/10536/DRO/DU:30062483
Chapter 15

Rehabilitation of Huntington’s disease

Belinda Bilney and Alan Pearce

Introduction

Huntington’s disease (HD) is an inherited neurodegenerative disease caused by an abnormally long cytosine-adenine-guanine (CAG) sequence on the 4p16.3 gene on the short arm of chromosome 4 [11]. The mutated CAG sequence leads to misfolding of the protein Huntingtin (Htt), which is widely expressed in the brain and other tissues of the body. Huntington’s disease is characterized by neuronal cell dysfunction and death, predominantly in the striatum. The disease is autosomal dominant with full penetrance, however age at disease onset is variable with around 50–70% of the variability positively related to the length of the abnormal CAG sequence [2]. The average age at disease onset is around 35–45 years and is confirmed by the presence of chorea, dystonia, bradykinesia, or rigidity in conjunction with a family history or positive gene test. Prodromal symptoms such as reduction in reaction and movement time, cognitive decline, and psychiatric distress are common [3] and may result in a reduction in participation in work, social, or leisure activities before observable disease onset.

The impairments associated with HD include a triad of motor, cognitive, and emotional disorders (see Table 15.1). Involuntary and voluntary movement disorders including chorea, dystonia, incoordination, bradykinesia, disruption of sequential and rapidly alternating movements, postural instability, gait disorders, and slowed voluntary eye movements may occur early in the disease; and rigidity, akinesia, hypokinesia (decrease in amplitude of movement), dysarthria, and dysphagia as the disease progresses. Cognitive decline, which often occurs early and is progressive, includes disorders of executive function, problem solving, attention, memory, verbal fluency, and comprehension [4–6]. Emotional changes characteristic of HD include depression, anxiety, irritability, and apathy. Irritability may present as perseverative thinking, which sometimes may result in anger when the person perceives their needs have not been met [7]. Apathy may in part be due to cognitive disorders associated with initiating, planning, sequencing, and completing a task [8]. Other symptoms associated with HD that may not be attributable to neural dysfunction include metabolic and endocrine disorders, weight loss, muscle wasting, and sleep disorders [9].

Impairments of motor, cognitive, and emotional function adversely affect the person’s ability to participate in family, social, and work activities. These impairments can be associated with difficulty maintaining employment, reduced social contact associated with communication and cognitive disorders, and cessation of sporting and recreational activities. As the disease progresses the person with HD may become more reliant on family for assistance with planning and completing a daily routine including personal and domestic activities of daily living.

Understanding the neuropathology associated with HD as a basis for developing therapeutic interventions

Huntington’s disease is thought to occur due to the mutant protein Huntingtin. Huntingtin is expressed within the brain and in other tissues outside of the central nervous system such as skeletal muscle [10]. The normal function of Huntingtin is still not well understood, although it appears to be necessary for normal development because mice with targeted disruption of Huntingtin do not survive beyond day 7 to 8 of embryogenesis [11]. In HD, there is a reduction of volume of the caudate, putamen, and white matter before manifest disease onset and further reductions


162
Table 15.1 Summary of rehabilitation needs through the progression of Huntington's disease

**Predominant Huntington's disease**
- Changes to body structure and function: decreased volume of caudate and putamen, white-matter atrophy
- Cognitive impairments: reduced processing speed, deficits of attention and working memory
- Emotional impairments: depression, irritability
- Motor impairments: reduced reaction time, bradykinesia of rapid alternating upper-limb movements and gait, reduced stride length, increased double-limb support and variability of stride length and step time, variability in grip force
- Activity limitations: driving, managing finances, decreased ability to supervise children, reluctance to use telephones, impaired social communication
- Participation restrictions: decreased ability to engage in usual work, decreased ability to volunteer, reduced participation in leisure activities

**Main goals of rehabilitation:**
- Preventive: establish healthy lifestyle; establish regular exercise program; strength and aerobic training; establish healthy diet
- Education: goal-specific education related to gene positive diagnosis; emotional support to cope with diagnosis
- Family counseling: regarding implications of gene positive diagnosis, emotional support
- Disciplines involved in rehabilitation: neurologist, genetics counselor, psychologist, psychiatrist, physiatrist, exercise physiologist, dietitian

**Early-stage Huntington's disease**
- Changes to body structure and function: loss of GABAergic medium spiny projections to globus pallidus and substantia nigra
- Cognitive impairments: comprehension of complex discourse, memory, planning, problem solving, organizing, new learning, attention
- Emotional impairments: depression, sadness, difficult to get along with, fatigue
- Motor impairments: chorea, postural instability and falls, reduced walking speed, reduced stride length and cadence, increased base of support, muscle weakness
- Activity limitations: postural instability during walking that may affect community ambulation, reduced accuracy of upper-limb function that may affect writing and manipulation tasks, difficulty with household chores and cooking
- Participation restrictions: reduced capacity for work

**Main goals of rehabilitation:**
- Preventive: prevent secondary impairments related to decreased activity
- Functional restoration: practices of communication skills, balance and gait retraining
- Education: goal fails prevention, strategies to maintain communication and social supports
- Family education: to support effective communication counseling to adjust to change in family dynamics and implications of diagnosis; dysphagia education
- Disciplines involved in rehabilitation: case manager/social worker; neurologist, neuropsychiatrist, general practitioner; rehabilitationist, physiatrist, speech pathologist, occupational therapist, dietitian, HD Association

**Mid-stage Huntington's disease**
- Changes to body structure and function: atrophy cerebral cortex, subcortical white matter, hypothalamic nucleus, muscle atrophy, endocrine changes, metabolic changes
- Cognitive impairments: slowed processing, impaired memory, inflexibility of thought, difficulty with sequencing, impaired attention
- Emotional impairments: anxiety, irritability, impatience, delusions
- Motor impairments: speech, dysarthric dysarthria; irregular articulatory breakdowns, prosodic changes, impaired phonation (harsh or strangled voice quality; volume variations)
- Movement: continued deterioration of postural instability and changes to gait, dystonia, chorea
- Activity limitations: may cease driving, community ambulation may become increasingly difficult
- Participation restrictions: may cease paid work, reduced participation in hobbies

**Main goals of rehabilitation:**
- Preventive cardiovascular and strength training program
Section 4 Rehabilitation of specific conditions

Table 15.1 (cont.)

Functional restoration: gait and balance training programs, task-specific practice of skills important to the patient
Compensatory approach: home modifications to aid independence and reduce risk of falls, strategies for enhancing memory and structuring daily routine, strategies to enhance communication, strategies to reduce risk of falls, strategies to aid swallowing, supplements to maintain healthy weight
Education goals: strategies for improving attention, managing memory deficits, structuring daily routines, decision-making about end-of-life care
Family: psychological support, practical support with equipment and aids, transport, home services, respite, financial support.
Disciplines involved in rehabilitation: as for early stages + home support services, respite care
Late stage Huntington’s disease
Changes to body structure and function: continued brain atrophy
Cognitive impairments: dementia
Emotional impairments: Cognitive Impairment may mask emotional Impairment
Motor impairments: rigidity, dystonia, hypokinesia, dysphagia, dysarthria, incontinence, muscle contracture, pain, chorea
Activity limitations: may no longer be able to walk independently, may require assistance for transfers (folded) and supported seating, assistance with wheelchair mobility
Participation restrictions: reduced participation in social and leisure activities because of communication, cognitive, physical impairment
Main goals of rehabilitation:
Preventative: maintain muscle length and joint range of motion to prevent pain and joint breakdown
Compensatory approach: modified seating, hoist for transfers, modified wheelchair, modified diet, modified mattress and bedding
Palliative approach: support for the process of dying, spiritual and emotional support, relaxation therapy, music therapy
Education goals: care-giver education for safe transfers and mobility, swallowing, chest care, contracture prevention / management and pressure care.
Family: ongoing psychological and financial support, assistance with care-giving at home or transition to residential or palliative care.
Disciplines involved in rehabilitation: as for mid-stages + residential care services, palliative care

In brain volumes including the globus pallidus, thalamus, and hypothalamus as the disease progresses. Microscopic studies show neuronal loss particularly of the gamma aminobutyric acid (GABA) enriched medium spiny neurons that project to the globus pallidus and substantia nigra. Reduced GABA and 5-hydroxytryptamine input to the external globus pallidus may cause hyperactivity of the external globus pallidus and inhibition of the subthalamic nucleus. In turn the excitatory drive to the major output nuclei of the basal ganglia is reduced, which may contribute to the hyperkinetic movement disorders associated with the early stages of HD. As HD progresses, the frontal and temporal areas that are connected to the pathological striatal circuits may atrophy and account for the cognitive and emotional impairments associated with the disease. The emerging evidence around the neuropathological basis of the motor, cognitive, and emotional disorders associated with HD can be used as a basis to design treatments including effective rehabilitation strategies.

Rehabilitation for people with HD
To date there is no known treatment that can delay or alter the progression of HD. There has been a large research effort examining the pathophysiology of HD with the hope of finding neuroprotective agents that may prevent or delay the onset of manifest disease [9]. There have also been reports of surgical interventions to attempt to ameliorate the symptoms of HD with deep-brain stimulation [12]. Stem-cell transplantation to support repair of the medium spiny neurons in the striatum might be possible in the future [13]. Although the efficacy of these types of treatments is still not known, it is likely that people who undergo surgical interventions will require intensive rehabilitation, including task-specific training, to maximize the benefit of the treatment [14].
A recent review of the European database of HD, which includes over 17,000 cases, showed that 57.9% of people with HD were prescribed medications for symptom management [15]. Trial data show that some medications are effective in ameliorating selected impairments associated with HD such as chorea, dystonia, depression, anxiety, irritability, and mania [16]. However, a recent Cochrane review showed there is no evidence to support the use of currently available pharmacology to modify the disease process [17].

Until there are significant advances that bring about the delay of disease onset or reduction in disease severity, the mainstay for the treatment of HD will continue to be a rehabilitation approach and, in the latter stages of the disease, care with a palliative focus. There has been very little attention in the scientific literature devoted to assessing the efficacy of rehabilitation therapies for people with HD [18, 19] and a framework for rehabilitation has not yet been established. Consequently, routine referral and participation in intensive rehabilitation programs is still not standard practice for people with HD [19–21]. It is possible that the triad of motor, cognition, and psychiatric impairments may lead some clinicians to believe that traditional approaches to rehabilitation, that require high levels of motivation and skill learning, may not be appropriate for people with HD [21]. However, there is emerging evidence that rehabilitation programs are effective in the short term for people with HD [21, 22] and greater research effort is required to determine the efficacy of specific therapies within a comprehensive rehabilitation program.

The goals of rehabilitation will vary between individuals and should be developed in consultation with the person with HD and if appropriate their family or caregivers. Possible goals of rehabilitation could include enhancing quality of life by facilitating independence in activities of daily living, and increasing or maintaining participation in work, social relationships, and leisure activities. Therapy goals could also be developed to reduce the risk of injury through falls and prevent secondary complications such as aspiration pneumonia, muscle contracture, pain, weight loss, and malnutrition. The effectiveness of rehabilitation for improving mood, and providing opportunity to form friendships and gain support from other people with HD and the rehabilitation team may also be important [22]. The onset and severity of the triad of impairments and consequent changes to activity and participation varies between individuals and changes across the disease duration.

Therefore, a rehabilitation model for people with HD needs to be flexible to accommodate these variations and also the coping capacity of individuals and their caregivers. Table 15.1 illustrates how the needs of the individual and their family changes throughout disease progression and how the composition of the treating team, treatment goals, and therapeutic interventions may also change to continue to meet the needs of the patient and their family [18, 23, 24].

### Family-centered rehabilitation for people with Huntington's disease

Rehabilitation of a person with HD may take a family-centered approach, which recognizes the critical role of the family in the care of the person with HD. This approach acknowledges that the family has an understanding of HD through their relationships with other family members diagnosed with the disease, and are therefore able to contribute to the rehabilitation plan, whilst respecting their needs, strengths, and ability to cope. A family-centered approach is important because of the substantial care provided by family members. For example, one study reported that carers provided an average of 6.9 hours care per day to dependents with HD [25]. This approach also recognizes that family members may provide care for an extended time, in some instances for the entire duration of the disease, which may be 15–20 years [25]. Many caregivers of people with HD report significant distress, anxiety, depression, and loss of independence associated with their care-giving role. This may be related to the high burden of care associated with caring for a person with impairments of memory, problem solving, initiative, depression, irritability, and apathy [20, 25, 26]. Dawson et al. [27] identified that both practical and psychological support may be required for the caregivers of people with HD. Practical support could be provided in the form of advice, provision of aids and equipment, and home modifications to maintain or improve the independence of the person with HD. Referrals could also be made to assist the family to access support within the home such as respite care, personal care, and home cleaning or maintenance. Psychological support may be offered to the family and person with HD throughout the disease duration. The rehabilitation team should have a good understanding of how a confirmed diagnosis of HD may affect the relationships within a family. For example, a person who has been diagnosed with HD may experience guilt or stress related to concern that
they may have passed on the disease to their children. They may also be in the position where they were providing care for a family member with HD but may no longer be able to do so because of their own symptom onset. Knowledge of how the disease has affected parents, siblings, or other close relatives may also contribute to anxiety about the disease. Furthermore, the roles of individuals within the family and effective communication between family members may be altered, particularly by the cognitive, emotional, and communication deficits associated with the disease.

**Composition of the rehabilitation team**

The range and complexity of symptoms associated with HD requires that over the duration of the disease a large team may work with the person and their family to minimize the activity and participation restrictions in work, family, and social life. The composition and size of the team may vary depending upon the stage of the disease and the emergence of motor, cognitive, and emotional impairments (see Figure 15.1). It is important that the patient’s management does not become fragmented [28], and for this reason it may be beneficial for rehabilitation to be coordinated using a case-management approach. The case-manager may be responsible for ensuring that well-timed referrals are made within the rehabilitation team so the person and their family receive accurate and timely information about HD across the disease continuum. Case-managers may also work to ensure that services to support the patient in their home and community are coordinated and align with the patient’s current rehabilitation goals. In some instances it may be appropriate for the case-manager to act as an advocate for the patient and their family within the rehabilitation team [19, 27, 29].
The challenge for rehabilitation teams is to ensure that all team members are knowledgeable about HD when the incidence of the disease and number of individuals referred to rehabilitation remains low. For example, a survey of 585 physical therapists showed that 15.5% had treated one patient with HD and only 6.2% had treated more than one person with the disease [30]. It is likely in the future, as has happened more recently for people with Parkinson’s disease, that more people with HD will be referred to movement rehabilitation and at an earlier stage of the disease progression [19]. This may require additional workforce education to ensure that the best evidence-based care is provided for people with HD.

Evidence related to effective models for rehabilitation

There is currently insufficient evidence to compare the efficacy of different models of rehabilitation, such as inpatient, outpatient, home-based, or longer-term maintenance programs. Zunzi et al. [21, 31, 32] reported that 3-week intensive blocks of planned inpatient rehabilitation conducted at a rehabilitation center in Italy were effective in improving function in the short term and maintaining function in people who continued to participate in the program for up to three times a year over 5 years [32]. They reported that the program included both individual and group therapy five and a half days a week for 3 hours a day. Rehabilitation included exercises to improve strength, aerobic fitness, flexibility, coordination, postural stability, and the efficiency of breathing and coughing. An adaptive approach was taken to improve functional tasks, including the practice of strategies to improve planning, concentration, memory, oral and written communication, and independence and safety in activities of daily living. The intensive program resulted in short-term improvements in function and mobility rated on the Physical Performance Test and the Timed Test [21], which was supported by the patient and care-giver perception of short-term improvements in domains such as balance, speed of movement, mood, apathy, and family and social relationships [31]. Although the efficacy of individual therapies within the intensive program cannot be determined, the patients and care-givers perceived the positive effects of the program to be related to an improved knowledge of HD and a greater sense of control in their disease management, opportunities to meet other people with HD, and the positive interactions with therapists [31].

Evidence related to the efficacy of outpatient or home-based models of rehabilitation is derived from single or multiple case studies to date [23, 33]. There is some evidence to suggest that less-intensive community-based rehabilitation that includes interventions such as strengthening, coordination, and balance exercises may also be effective in the short term, but clearly more evidence is required to determine if improvements in function are maintained and which elements of the intervention are effective. Therapy that is individualized but conducted within a group setting such as balance retraining, gym-based strength and aerobic training, and communication or memory training groups may be more economical ways to provide ongoing rehabilitation. However, there is currently no evidence that evaluates this model of rehabilitation for people with HD.

Rehabilitation across the disease continuum

Prodromal phase

Many people who are gene positive for HD but have not yet been diagnosed with manifest disease onset report changes in activity including difficulties with driving, social communication, managing finances, completing household tasks, and shopping [34]. Symptoms including cognitive impairment, depression, bradykinesia and rigidity in force production, and difficulty with fine manipulation [35–37]. Gait changes including reduction in speed and stride length may also be noted [35] (see Table 15.1). Close to the time of disease onset, the team involved in care might include a neurologist, genetic counselor, social worker, or psychologist. Timely, accurate, and individualized information needs to be provided about HD around the time of diagnosis, with care to ensure that not too much information is given, which has been found to be stressful for people with HD [27]. Goals of intervention in the prodromal stage may include establishing a healthy lifestyle through diet and exercise.

The understanding of the efficacy of exercise, either to prevent or delay disease progression or as a tool for rehabilitation in HD, is currently limited. Recently there has been some evidence from mice models that exercise, within an "enriched" environment, may prevent the loss of cerebral volume and
delay the onset of HD [38-40]. Collectively the studies used transgenic mice, bred to develop the features of human HD, randomly allocated into two groups: one group were provided in their home cage objects to "enrich" the environment (such as cardboard boxes, open wooden boxes, cylindrical tunnels, and other non-toxic plastic objects); whilst the control group were housed in home cages with only wood shaving for bedding. Results showed that in all studies the mice exposed to an enriched environment demonstrated a delay in the onset of HD symptoms compared to the control mice.

These findings have been cited as evidence to support the use of exercise and cognitive challenges, either as a preventative measure, or as a rehabilitation strategy to impede disease progression. As with any research involving animal models, translating the findings to humans should be viewed with caution. Kosinski et al. [41] presented a case study of a 38-year-old person with HD who trained seriously as a marathon runner from the years of 1989 to 1997, with an average completion time of 2 hours and 40 minutes (competitive semi-professional running standard). In 1998, the patient started to experience severe muscle fatigue and pain after running, with completion times gradually declining to around 3 hours and 20 minutes. The patient was initially diagnosed with mild myopathy and mitochondrial pathology, but was also diagnosed with HD in 2004. Although the study presented by Kosinski et al. [41] focused on myopathy as a presenting early symptom of HD, in a review of the original case Aitschuler [42] suggested that exercise did not appear to delay the development of HD in this patient whose disease onset was 20 years earlier than the median age at onset. Moreover, although Aitschuler [43] suggests that exercise may be the most transferable factor from the mice studies to humans, the environment provided to stimulate the mice may not be applicable for humans. In other words, the studies showing positive signs in mice may be too simplistic for the environment humans currently live in.

Although clear evidence is lacking for the prescription of exercise for people with HD, there is still a possibility that exercise may delay the onset and/or progression of HD [42]. It may be via an optimal amount of exercise (rather than extremes in the case of the marathon runner). As suggested by Bilney et al. [18] and Busse and Rosser [14] there is a small amount of evidence demonstrating that exercise may be helpful for range of movement and balance decrements for people with HD. For example, Quinn and Rao [33] presented a case study showing that after 14 weeks of a home exercise program involving movement to music, and functional and balance activities, the patient's SF-36, Berg Balance Scale, and Unified Huntington's Disease Rating Scale (UHDRS) showed noticeable improvements.

Early stages of HD

The presentation of involuntary movement disorders may signal disease onset together with depression, sadness, difficulty to get along with people, lack of motivation, and fatigue [43]. There may be progression of cognitive deficits including impairment of memory, planning, problem solving, organizing, new learning, and attention. Subtle language and comprehension disorders may also be present [4]. Movement disorders may include the onset of mild postural instability [44], increased bradykinesia of gait, and reduction in quality of hand writing due to chorea, bradykinesia, difficulties with movement sequencing, and variability in grip force [45]. Rehabilitation-team goals include: completion of speech and language, occupational therapy, and physiotherapy assessments with the purpose of determining baseline function; early intervention to maintain participation in work, driving, and social activities; and development of relationships between the team, the patient, and their family [23]. Strategies may need to be put in place to provide psychological support for the person with HD and their family, and individualized education may also be required.

Initial speech-pathology assessment may include evaluation of communication initiative, intelligibility, word-finding ability, and verbal fluency. Deficits of comprehension of complex discourse may also be present in early disease and may be assessed so that strategies can be put in place to improve communication, including education of family members on how to be supportive communication partners [46].

Occupational therapy assessment of functional tasks including work, driving, leisure, domestic, and personal activities of daily living could include evaluation of how the movement disorders in combination with cognitive disorders, reduced motivation, and fatigue may affect function. Assessment of work capacity and the development of strategies to assist in maintaining work capacity or negotiation to modify...
work tasks may assist in prolonging paid employment. An off- and on-road driving assessment may be required to determine the person's capacity to drive and to make recommendations for retraining or restrictions to driving conditions [47]. An early home visit may also be required to provide advice on environmental adaptations or equipment that may improve safety or increase independence in the home. A home visit may also provide an opportunity to identify activities that are important to the person with HD that could be improved through task-specific practice.

Physiotherapy intervention may include support for maintenance of an independent exercise program and intervention to reduce falls risk and improve postural stability.

A previous systematic review [18] suggests exercise rehabilitation for people with HD is usually directed at symptomatic management of the primary movement disorders and prevention of secondary impairments related to the primary movement disorder and immobility. In particular, the efficacy of exercise therapy to improve cardiovascular fitness, muscle strength, and flexibility has been tested [48, 49]. Studies include the evaluation of nat exercises [30, 48], seated strengthening exercises [50], strengthening and cardiovascular exercises [51], and hydrotherapy [52]. However, the levels of evidence for studies included in this review were classified between levels IV (observational studies), V (non-experimental studies), VI (expert opinion); thereby demonstrating the published evidence was not strong [18, 50].

Recent falls can be a significant problem for people with HD and may occur in the early and middle stages of HD [53, 54]. Retrospective reporting of the annual incidence of recurrent falls (two or more) in the early and middle stages of HD is around 58.3–60% [54, 55] or 36% over a six-month period [56]. The ability of people with HD to accurately report their fall frequency and injuries associated with falls decreases with decline in cognitive status [53] and therefore prospective studies and care-giver reporting are likely to provide a more accurate picture of falls frequency. Unfortunately there is limited prospective reporting of falls in the literature; however, one study reported that 40% of 45 participants fell once and 20% fell twice over a three-month period [54].

The cause of falls in people with HD is most likely to be multi-factorial and a large number of impairments that could contribute to postural instability have been identified in the literature. These include problems with correct sequencing of postural responses, delays in initiation and prolonged anticipatory responses [44, 57, 58], deficits in adapting the postural response to changing task demands or environment including response to chorea, increased postural sway [54], altered musculoskeletal alignment which may be associated with chorea and dystonia, disorders of sensory information integration including dependence on proprioceptive cues more than visual cues [44], impaired cognitive function [53, 54], reduced ability to maintain postural stability during dual tasks [54], lower-limb muscle weakness [59], reduced activity [55], and behavioral disturbances [54]. The side effects of some medications and environmental hazards may also contribute to the high incidence of falls.

Given the large number of impairments that could be contributing to postural instability in people with HD, a comprehensive assessment of balance and falls may be required so that interventions can be targeted to address the primary impairment(s). Interventions could include using an attentional strategy to prepare anticipatory responses prior to commencing a task and to identify potential threats to stability, and retraining the stepping strategy to improve magnitude and timing of the step response to perturbation. Lower-limb muscle strengthening, task-specific balance retraining [23], environmental modification to reduce risk of falls, and medication review may also be effective in improving instability and reducing falls. As suggested, further studies with improved research designs and larger sample sizes are required to ascertain the effectiveness of exercise therapy for people with HD. Moreover, it is likely that effective therapy for HD is likely to be multifaceted, combining exercise with cognitive therapies.

Mid stages of HD
As the disease progresses people with HD may find it difficult to continue with full-time employment due to problems with memory, slowed processing, impaired comprehension, inflexibility of thought, and fatigue. Driving may no longer be possible due to slowed reaction time, difficulty maintaining attention, and slowed processing. The person with HD may find it increasingly difficult to manage household tasks due to postural instability and bradykinesia. Community access for shopping and leisure may also become
more difficult due to reduced gait speed and postural instability.

The gait disorder in HD includes reduction in speed due to decreased step length and cadence, increased base of support, and increased variability of step length, swing and double-limb support time [35, 51, 60]. Kinematic changes include reduction in the hip and knee angle excursions in the sagittal plane, increased lateral sway, and reduced arm swing [58]. People with HD may have difficulty turning, deterioration of gait during dual tasks [61], and delays in gait initiation (akinesia) [58]. Progressive gait deterioration may result in loss of independence in walking, which is a predictive factor for admission to residential care.

Gait retraining strategies could include training people with HD to use attentional strategies to reduce akinesia, improve postural stability during mobility, and reduce the frequency of falls [61]. Training people with HD to effectively allocate their attention between two tasks when walking may also be effective. If retraining for flexibility in allocation of attention is not possible, particularly as the cognitive deficit progresses, teaching people to compensate by avoiding dual tasks may be a more effective approach. Several studies have examined whether external auditory cues can be used to modulate walking by bypassing the defective basal ganglia [51, 52, 63]. Unlike people with Parkinson's disease, people with HD have difficulty synchronizing their stepping to a cue [51], and there is inconsistency in the literature as to whether the cues result in short-term improvements in cadence, step, and stride length [51, 62, 63]. Delval et al. [62] suggests that external cues may not be of assistance because of the attentional deficits characteristic of HD. The prescription of gait aids may not be useful for people with HD because of the difficulty with allocating attention to a dual task and difficulty controlling the gait aid if chorea is severe [23]. Similarly, few people with HD are able to use an electric wheelchair or motorized scooter safely because of difficulty with attention, fine motor control, and bradykinesia [23].

Late stages of HD

The late stages of HD may be characterized by the progression of cognitive impairments resulting in dementia, dysphagia, dyskinesia, and incontinence. Communication may become more difficult due to the progression of dysarthria and dementia. Secondary complications may include muscle contracture, pain, and skin breakdowns due to abnormal positioning associated with dystonia and rigidity, and aspiration pneumonia and weight-loss associated with dysphagia. The focus of intervention in this phase may move from a restorative to a preventative, compensatory, or palliative approach. The physiotherapist, nurse, occupational therapist, speech pathologist, and diettician may continue to work together to attain positioning that promotes safe swallowing, prevents secondary impairments of muscle contracture and skin breakdown, and encourages communication. Specialized seating, wheelchairs, and hoists for transfers may need to be prescribed in the late stages of the disease. Carers are often trained to provide passive joint range of motion and muscle-stretching exercises to maintain range of motion; however, the efficacy of this treatment for people with HD is not known. Airway-clearance techniques such as deep breathing, postural positioning, and supported coughing are often used and can be taught to the family and other staff who may be caring for the person with HD.

In the late stages of the disease, carers may find that they cannot provide the required care at home and residential care may be required. Predictors for requiring institutional care include reduced capacity to complete activities of daily living and poor motor function [64]. There are few specialized residential care facilities for people with HD [27] and in some cases there may be a reluctance to accept people with HD, which may be due to a limited understanding of the disease and a perception that the care requirements are very high due to the cognitive, emotional, and movement disorders [65]. As the person with HD transitions to supported care, the rehabilitation team may be involved in educating the residential care team and in providing ongoing advice and assistance for the patient, family, and professional team.

Conclusion

Although the specific goals of rehabilitation for individuals with HD will vary, they may include enhancing quality of life by maintaining independence, reducing the risk of injury through falls, and by prevention of secondary complications related to the movement disorder. Rehabilitation goals may be achieved through the specific practice of tasks such as activities of daily living and walking, exercises to maintain strength, range of movement, and cardiovascular fitness, practice of cognitive tasks or the implementation of
compensatory strategies to enhance cognitive function, and modification of the environment to maintain independence or reduce risk of falls. Despite a lack of clear evidence for the prescription of exercise, observational and case-study data reveals that rehabilitation involving functional tasks, balance and posture, and breathing exercises may delay the progression of HD. There is emerging evidence that multidisciplinary and intensive rehabilitation programs may have some short-term benefits for people with HD in the physical, cognitive, and emotional domains; however, the effectiveness of individual therapies within comprehensive rehabilitation programs is not yet known. Further research is required to ascertain the benefit of cognitive, occupational, and exercise therapy for people with HD. Although there is currently insufficient evidence on the efficacy of different models of rehabilitation, it is possible that therapy based upon a family-centered approach may reduce the stress sometimes experienced by the primary caregivers of a person with HD. Until there are significant advances in the capacity to delay the onset or reduce the severity of the disease, therapy provided by a knowledgeable and multidisciplinary team may be best placed to provide a coordinated and responsive therapy approach to meet the needs of the person with HD and their family throughout the disease continuum.

References

Section 4 Rehabilitation of specific conditions


Chapter 15: Rehabilitation of Huntington's disease


Movement disorders affect a growing patient population, but providing comprehensive care is extremely difficult. Several of these conditions are progressive and incurable; the basal ganglia have a complex role in movement control, with many potential malfunctions. This book focuses on rehabilitation approaches that have been developed and utilized internationally in an attempt to minimize impairment and maximize participation amongst these patients.

Each chapter is written by movement disorders experts, rehabilitation specialists, and healthcare professionals, giving a broad overview of current interventions, and emphasizing the need for interdisciplinary management, focusing on deliverable outcomes. Common conditions such as Parkinson’s disease, cerebral palsy, dystonia, and Huntington’s disease are comprehensively covered. This book gives neurologists, geriatricians, and rehabilitation specialists an up-to-date, theoretically based approach to managing movement disorders related to basal ganglia malfunction. Also valuable for physiotherapists, occupational therapists, speech pathologists, nurses, and social workers seeking to develop and plan appropriate interventions.

Other titles of interest:

Uncommon Causes of Movement Disorders
Edited by Nestor Gálvez-Jiménez and Paul Tuite
(ISBN: 9780521111546)

Tremor, Parkinson’s Disease and Related Movement Disorders
Edited by Peter LeWitt
(ISBN: 9780521140442)

Neuropsychiatric and Cognitive Changes in Parkinson’s Disease and Related Movement Disorders
Edited by Dag Aasland, Jeffrey Cummings, Daniel Weintraub, and Ray Chaudhuri
(ISBN: 9781107039223)
Contributors

Giovanni Abbruzzese, MD
Director, Clinica Neurologica 2, Department of Neurosciences, Ophthalmology and Genetics, University of Genoa, Italy

Brooke Adair, BPhys(Hons)
Department of Physiotherapy, University of Melbourne, Parkville Australia

Ana Aragon, Dip. COT
Independent Occupational Therapist and Associate Senior Lecturer, Faculty of Health and Social Sciences, Leeds Metropolitan University, Leeds, UK

Alfredo Berardelli, MD, PhD
Professor of Neurology, Department of Neurology and Psychiatry and NeuroMed Institute, Sapienza University of Rome, Rome, Italy

Belinda Bilney, BAppSci (Physio), PhD
Lecturer, Melbourne School of Health Sciences, Department of Physiotherapy, University of Melbourne, Australia

David J. Brooks, MD, DSc, FRCP, FMedSci
Harrnett Professor of Neurology and Head, Centre for Neuroscience, Department of Medicine, Imperial College London, Hammersmith Hospital, London, UK

Emma Campagna, BSc
Friedreich Ataxia Clinic, Department of Physiotherapy, Monash Medical Centre, Southern Health, Clayton, Victoria, Australia

Louise A. Corben, BAppSci (OT), MSc, PhD
Bruce Lefroy Centre, Murdoch Children's Research Institute, Parkville; and Friedreich Ataxia Clinic and Occupational Therapy Department, Monash Medical Centre, Southern Health, Clayton, Victoria, Australia

Mary Danoudis, Dip Physiotherapy; BAppSci (Physio); Masters Physiotherapy
Clinical Research Centre for Movement Disorders and Gait, Kingston Centre, Cheltenham, Victoria, Australia

Martin B. Delatycki, PhD
Professor, Bruce Lefroy Centre, Murdoch Children's Research Institute, Parkville; Clinical Genetics, Austin Health, Heidelberg, Victoria; and Friedreich Ataxia Clinic, Monash Medical Centre, Southern Health, Clayton, Victoria, Australia

Georg Dirnberger, MD, PhD, MSc
Specialist in Neurology, Visiting Researcher, Department of Neurobiology, Weizmann Institute of Science, Rehovot, Israel

H. Kerr Graham, MD, FRCS (Ed), FRACS
Professor of Orthopaedic Surgery, University of Melbourne, Royal Children's Hospital, Melbourne, Victoria, Australia

Ralph Hampson, PhD
Director, Ralph Hampson Consulting, Victoria, Australia

Robert Iancek, BMedSci, MBBS, PhD, FRACP
Director, Clinical Research Centre for Movement Disorders and Gait, Kingston Centre, Southern Health, Victoria, Australia

Marjan Jahanshahi, BSc, Mphil (Clin Psychol), PhD
Professor of Neuropsychology, Sobell Department of Motor Neuroscience and Movement Disorders, Institute of Neurology, University College London; and the National Hospital for Neurology and Neurosurgery, London, UK
Lynette Joubert, BA, BSocSci, MA Clin Psych, D Litt et Phil
Associate Professor, University of Melbourne Department of Social Work, Victoria, Australia

Jill Kings, Dip. COT, MSc (Neuro Rehab)
Senior Lecturer, Allied Health Continuing Professional Development, Faculty of Health and Social Sciences, Northumbria University, Newcastle-Upon-Tyne, UK

Sue Lord, PhD
NIHR Research Fellow, Clinical Ageing Research Unit, Institute for Ageing and Health, Newcastle University, Newcastle-Upon-Tyne, UK

Andres M. Lozano, MD, PhD, FRSC, FACS
RR Tasker Chair in Functional Neurosurgery, Division of Neurosurgery, Toronto Western Hospital, Toronto, ON, Canada

Victor McConvey, RN, MACN
Parkinson’s Nurse Consultant, Parkinson’s Victoria, Cheltenham, Victoria, Australia

Rachael McDonald, BAAppSc (OT), PGDip (Biomech), GCHE, PhD
Senior Lecturer, Centre for Developmental Disability Health, Monash University, Victoria, Australia

Jennifer L. McInley, BA App Sc (Physio), Grad.Dip. Neurosciences, PhD
Senior Lecturer, Department of Physiotherapy, The University of Melbourne, Australia

Kalthida Methawasin, MD
Department of Neurology, Parkinson’s Disease and Movement Disorders Centre, National Neuroscience Institute, Singapore

Sarah Milne, BPhysio
Physiotherapy Department, Kingston Centre, Southern Health, Cheltenham, Victoria, Australia

Meg E. Morris, BAAppSc (Physio), Grad Dip Gerontology, MAAppSc, PhD, FACP
Professor, School of Allied Health, Faculty of Health Sciences, La Trobe University, Bundoora, Australia

John Olver, MBBS, MD, FACRM, FAFRM
Victor Smorgon Chair of Rehabilitation Medicine, Faculty of Medicine, Nursing and Health Sciences, Monash University, Victoria, Australia

Nicola Pavesse, MD
Clinical Senior Lecturer in Neurology, Centre for Neuroscience, Department of Medicine, Imperial College London, Hammersmith Hospital, London, UK

Alan Pearce, BSoc (Hons), Grad Dip (Ex Sci), PhD
Director, Cognitive and Exercise Neuroscience Unit, School of Psychology, Deakin University, Burwood, Australia

E. Diane Playford, MD, MRCP, MBBS
Reader In Neurological Rehabilitation, Institute of Neurology, Faculty of Brain Sciences, University College London, London, UK

Barry Rawick, MBBS, FACRM, FAFRM (RACP)
Associate Professor, Department of Medicine, Monash University, Victoria, Australia

Nicole Rinehart, BA, M Clin Psych, PhD
Associate Professor, Centre for Developmental Psychiatry and Psychology, School of Psychology & Psychiatry, Monash University, Victoria, Australia

Lynn Rochester, PhD, Grad Dip Phys
Professor of Human Movement Science, Clinical Ageing Research Unit, Institute for Ageing and Health, Newcastle University, Newcastle-Upon-Tyne, UK

Chloe Stanley-Cary, BA, DPsych (Neuro)
Centre for Developmental Psychiatry and Psychology, School of Psychology & Psychiatry, Monash University, Victoria, Australia

Antonio Suppa, MD, PhD
Post Doctoral Research Fellow, Department of Neurology and Psychiatry and NeuroMed Institute, Sapienza University of Rome, Rome, Italy

Louis C. S. Tan, MBBS, MRCP, FAMS, FRCP
Senior Consultant, Department of Neurology, Parkinson’s Disease and Movement Disorders Centre, National Neuroscience Institute, Singapore
Index

Spilberger Stait-Trait Anxiety Inventory, 59
spinal cord, 19, 20, 47-9, 135, 136, 185, 188
spinal-cord injury, 193, 195
spino-cerebellar ataxia, 25, 153
sport, 215, 222
status dystonicus, 31
Steele-Richardson-Olszewski syndrome, 152
stereotactic surgery, 5
sport, 215, 222
spinal-cord injury, 192, 195
spinocerebellar atrophy, 48
striatal degeneration in Huntington’s disease, 8-11
stroke
dystonias associated with, 45
Stroop test, 98, 100
substantia nigra, 1-2
substantia nigra pars compacta, 14
substantia nigra pars reticulata, 14
subthalamic nucleus, 1-3, 14
Sudek’s atrophy, 48
supervisory attention system (SAS) concept, 95
supplementary motor area, 2-7
surgical interventions
delay in movement disorder management, 36
See also functional neurosurgery, peripheral surgery; single event multilevel surgery (SIRM/S).
swallowing disorders, 116-117
assessment, 117
management, 117-118
swallowing management and secretion management, 85-86
Friedreich ataxia, 195
synuclein immunocytochemistry, 4
tai chi, 134
Tardieu Scale, 190
tardive dystonias, 111, 112
tardive dystonia, 40
task-specific dystonias
botulinum toxin treatment, 45-46
task-specific kinetic tremor, 30
tau protein pathology
cortical basal degeneration, 155
progressive supranuclear palsy, 152
technology
role in speech disorder rehabilitation, 118-119
telemetabolism, 118
temporal processing, 103
Test of Language Competence – Expanded Edition (TLCE-E), 116
tetrahydrozine, 33, 174
thalamotomy, 31, 39
thalami, 1, 2, 3
The Word Test – Revised (TWT-R), 116
thermal regulation disturbance, 87
tic disorders, 82, 111, 112
DSM-IV-TR criteria, 217
tics, 35
Timed Up and Go test (TUG), 83, 236
Tinetti scale, 167
tocclavus, 28
topiramate, 30, 39
torsion dystonia, 48
Tower of London test, 7, 98, 100
Trail Making test, 98
transcranial magnetic stimulation (TMS) studies dystonia, 17-18
Gilles de la Tourette syndrome, 19
Huntington’s disease, 19
Parkinson’s disease, 15-16
transfer movements
physiotherapy interventions, 64
traumatic brain injury, 194
rehabilitation principles, 131-133
tremor, 25, 82, 111
action tremor, 30
botulinum toxin treatment, 46-47
cerebellar tremor, 30
classification, 30
clinical evaluation, 30
definition, 30
esential tremor treatment, 30-31
in Parkinson’s disease, 14
intention tremor, 30
isometric tremor, 30
kinetic tremor, 30
medical management, 30-31
position-specific postural tremor, 30
postural tremor, 30
rest tremor, 30
task-specific kinetic tremor, 30
treatment, 30-31
trihexyphenidyl, 26, 39
Unified Huntington’s disease Rating Scale (UHDRS), 113, 168
Unified Parkinson’s disease Rating Scale (UPDRS), 6, 83, 147, 148, 235
upper- and lower-limb function 157
parkinsonian syndromes, 157
upper-limb dysfunction
physiotherapy interventions, 64
urinary-tract infections, 89
utilization behaviors, 103
validity of outcome measurements, 236
vascular parkinsonism, 35, 152, 153
dysphagia management, 157-158
gait and mobility rehabilitation: issues, 156-157
home and environmental assessment, 157
medical management, 158-159
multidisciplinary team care, 159
nursing and community care, 158
speech and communication difficulties, 157
therapeutic strategies, 156
upper- and lower-limb functional issues, 157
verbal fluency tests, 100
vestibular dysfunction, 82
visual problems
in Friedreich ataxia, 187
visuo-spatial function tests, 98
tonic tics, 112
voice tremor, 113
wearing-off effect with levodopa, 28
Wechsler Adult Intelligence Scales (WAIS-III or IV), 98
WEBMOVE web page, 241
Western Aphasia Battery – Revised (WAB-R), 116
Wilg-Semel Test of Linguistic Concepts (WSTLC), 116
Wilson’s disease, 25, 153
Wisconsin card sorting test, 98, 100
working memory model (Baddeley), 95
working memory tests, 98
writer’s cramp, 39, 45
botulinum toxin treatment, 45-46
rehabilitation strategies, 177-178
yoga, 134
zonisamide, 30
Zung Self-rating Anxiety Scale, 99