PHYSIOTHERAPY BRIEFING

Huntington's disease

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Early physiotherapy intervention plays a key role in management of Huntington's Disease (HD) over the life cycle of the disease.

What is Huntington's disease?

HD is an inherited neurodegenerative disease for which there is no cure. Children of an affected individual have a 50/50 chance of inheriting the faulty HD gene.

The mutation results in degeneration of the basal ganglia. This is a structure in the brain that has links to most other brain areas and is involved in coordination of body movment. People with HD gradually develop physical, cognitive and behavioural difficulties. Ability to maintain functional independence becomes limited by involuntary movements, altered muscle tone, impaired balance, and impaired speech and swallowing. Difficulties with memory, planning, prioritising and problem solving as well as behavioural issues pose an additional challenge for people with HD.

Appearance of symptoms varies between individuals but it generally affects individuals between the ages of 35-60 years of age [1]. Following formal diagnosis, symptoms progress over 17-20 years until full nursing care is required. Balance and mobility problems are a common feature [2], leading to falls, decreased walking ability and sedentary lifestyles [3].

How can physiotherapy help?

People with HD require ongoing health and social care to support their changing needs [4]. Maintaining fitness and physical function from early on in the disease process is vital to maintain quality of life in HD and is especially important given the duration of the disease and known burden on healthcare services. Ongoing efforts to define physiotherapy interventions [5], validate outcome measures [6] and conduct robust evaluations of targeted physiotherapy led interventions [7–10] have shown that people with HD can achieve fitness and measurable functional benefit from specific exercise training. Indeed community and home based exercise interventions have also been shown to be enjoyable, acceptable and beneficial for people with HD.

In addition, with referrals from physiotherapists, people with HD can access exercise support in local gyms through exercise referral schemes (for example the National Exercise Referral Scheme (NERS) in Wales)[7,8,11]. In the later stages of the disease multi-disciplinary rehabilitation, incorporating physical, occupational therapy, speech therapy and social activities has potential to moderate disease progression has been shown to be crucial to maintaining function [12-14].

Physiotherapy should focus on maintaining fitness, mobility and function and includes:

- Early access to evidence based self-management support to establish strategies for regular sustained physical activity [10] to minimise effects of primary and secondary impairments, including weakness, muscle imbalances, balance impairments and falls.
- Signposting to local support via the Huntington's disease Association of England and Wales (or Scottish Huntington's Association).
- Assistance with managing the altered walking patterns that often develop across the life cycle of disease including advice on carrying out activities of functional mobility, walking aids [15], footwear and discussing adaptations in the home for optimal safety.
- Monitoring of respiratory function from the early stages of the disease to limit the potential for developing respiratory complications [17].
- Encouraging independent walking for as long as is reasonably possible given the high risk of complications due to inactivity and sedentary behaviour.
- Advise carers, nursing home staff and other allied healthcare professionals on positioning and respiratory care in the later stages of the disease.

Case study:

An intervention approach that promotes autonomy, competence and relatedness can improve self-efficacy for secondary prevention to slow or alter disease progression in HD. A recently completed multi-centre (8 sites) trial (ISRCTN 65378754) of an individualised life-style approach to enhancing physical activity in HD compared to social interactions resulted in Life Space scores (i.e. a measure that quantified the extent of movement within a person's environment) that were 16 points higher in the physical activity group than in the social group [95% CI 2,30]. Self-reported Physical Activity (IPAQ) scores were also 125% higher in the physical activity group [95% CI: 4%, 388%] and exercise self-efficacy was 1.63 points higher [95% CI 0.48, 2.78].

"Early physiotherapy intervention in HD is essential to prolong fitness, mobility and function "

Size of the problem:

- Approximately 12 in every 100,000 people in the United Kingdom (UK)[18] are affected by HD.
- HD is dominantly inherited meaning that every person who inherits the gene will develop symptoms.
- Each child of a person with HD has a 50% chance of inheriting the faulty gene
- From formal diagnosis, duration of HD is approximately 17-20 years [17].
- Aspiration pneumonia is thought to be the leading cause of death in HD [19].

Conclusion:

Physiotherapy has an important role in encouraging and supporting people with HD to engage in activities to maintain their health, functional abilities and quality of life from early on in the disease process. Physiotherapists should be aware of the need to adapt their approach in tailoring to the individual needs of the person with HD and carer(s) who support them.

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For more information see:
www.activehd.co.uk and http://hda.org.uk/

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