

Institution: Cardiff University		
Unit of Assessment: Allied Health Professions, Dentistry, Nursing and Pharmacy (3)		
Title of case study: New physiotherapy standards and guidelines to improve management of Huntington's disease		
Period when the underpinning research was undertaken: 2007 - 2013		
Details of staff conducting the underpinning research from the submitting unit:		
Name(s):	Role(s) (e.g. job title):	Period(s) employed by submitting HEI:
Monica Busse	Professor	01/03/2005 - present
Anne Rosser	Professor	06/01/2003 - present
Lori Quinn	Senior Research Fellow	28/06/2002 - 12/06/2006
Una Jones	Senior Lecturer	01/08/1998 - present
Karen Jones	Senior Lecturer	01/07/2004 - present
Period when the claimed impact occurred: 2013 - 2020		
Is this case study continued from a case study submitted in 2014? No		
1. Summary of the impact (indicative maximum 100 words) Huntington's Disease (HD) is a neurodegenerative disease with no curative treatment options. Research by Cardiff's Huntington's Disease Group provided the first quantitative evidence that physiotherapy and regular supported physical activity were beneficial for people with HD. The findings were the catalyst for the first physiotherapy guidelines for HD, and for the subsequent inclusion of physiotherapy as a key intervention in HD management guidelines used across eight European nations and internationally. From 2015, Cardiff researchers also supported the annual accredited course to UK HD health professionals (via the HD Association), as well as development of patient resources now used internationally (e.g. by the Chinese HD Association). These were particularly vital during the COVID pandemic when many HD clinical services were paused.		
2. Underpinning research (indicative maximum 500 words) Huntington's Disease (HD) is a progressive neurological disorder that causes uncontrolled movements, loss of cognitive ability, as well as irritability, depression and other mood changes. The most common monogenetic, neurodegenerative disease worldwide, it affects 12 people in every 100,000. The personal, social and economic consequences of HD are devastating; for example, the cost of treating the 9,000 people with HD in the UK equates to £195M every year. Prior to Cardiff research, there was no standard guidance for the use of physiotherapy, including home exercise, in HD treatment. Consequently, physiotherapists working with HD patients primarily relied on clinical judgement rather than evidence-informed guidelines for treatment planning. Professor Ed Wild, Associate Director of the UCL Huntington's Disease Centre confirms: <i>"most community physiotherapy teams...[had] little to no experience of the very specific and unusual physical and cognitive impairments that HD patients can display"</i> with many clinicians tending to <i>"underestimate the potential benefits of physiotherapy in these patients"</i> [5.1a]. To address this knowledge gap, Busse and Rosser undertook a series of research studies, randomized control trials, and a systematic review designed to assess the efficacy of physiotherapy interventions for people with HD, and whether those delivered positive physical, cognitive and / or social benefits. 2.1 Physiotherapy as an intervention in HD: Busse and Rosser sought to characterise physiotherapy practice for people with HD in order to develop a standardised framework for		

patient management that could support clinical care. Through a mixed methods study, the research found that [3.1]:

- physiotherapy outcome measures were underused to assess efficacy of care;
- physiotherapy itself was underused, particularly in the early stages of the disease;
- management of falls and mobility deficit progression is core to treatment of people with HD.

This research supported development of a new consensus framework for physiotherapy in the care of HD patients, importantly highlighting that clinical practice should be developed according to the disease stage of progression [3.1].

2.2 Outcome measures for physical functioning: Having highlighted the importance of falls management as a preventive treatment goal, the team also carried out a global validation study for HD performance-based outcome measures [G3.1], assessing 11 items (e.g., 6 Minute Walk Test distance, Physical Performance Test (PPT), Barthel Index) across different stages of disease progression. The research identified that the Berg Balance Scale (BBS) and Timed “Up & Go Test” (TUG) were the most appropriate tools for practitioner assessment of a patient’s risk of falling [3.2]. They also found that the PPT had excellent utility as an effective outcome measure of physical ability. These three tests were highlighted as appropriate measures for clinical trials testing interventions focused on improving physical functioning in people with HD [3.2].

2.3 Exercise as an intervention: In 2012, the Cardiff team conducted the first ever randomized controlled trial (RCT) of a physiotherapy intervention in patients with early to mid-stage HD [3.3]. The intervention (a home exercise DVD, *Move to Exercise*) improved movement outcomes (e.g., gait speed, balance, function and level of activity) in HD patients, compared to a control group. Building on this work, in 2013, the researchers undertook a further randomized control trial into a gym-based exercise intervention (supplemented by a home walking programme) [3.4, G3.2]. The trial demonstrated improvements in participants’ physical ability, as well as in their mental health [3.4]. The study was awarded the *Journal of Neurological Physical Therapy*’s prize for the best publication of 2013, based on its contribution to knowledge, design and implementation of physiotherapy practice. In 2013, the team used the findings from these two trials to develop the ENGAGE-HD intervention and workbook (revised in 2020) [G3.3]. These trials showed that a physiotherapy intervention was feasible, and coaching could improve motivation, physical activity, and cognition in people with HD [3.5].

2.4 Systematic review of HD and physiotherapy: A subsequent systematic review of evidence-based clinical management of HD using physiotherapy, based on 18 studies, found that physiotherapy interventions are of benefit to HD patients, both in terms of physical and mental health, but that heterogeneity of outcome measures used across studies remained a limitation to drawing comparative conclusions from the various studies [3.6].

3. References to the research (indicative maximum of six references)

- [3.1] Busse M, Khalil H, Quinn L, Rosser A. Physical therapy intervention for patients with Huntington’s Disease. *Physical Therapy*. 2008, 88 (7): 820-831. DOI: 10.2522/ptj.20070346
- [3.2] Quinn L, Khalil H, Dawes H, Fritz N, Kegelmeyer D, Kloos A, Jonathan Gillard, Busse M, and the Physiotherapy Working Group of the European Huntington’s Disease Network. Reliability and minimal detectable change of physical performance measures in individuals with premanifest and manifest Huntington’s disease. *Physical Therapy*. 2013, 93 (7): 942-956. DOI: 10.2522/ptj.20130032
- [3.3] Khalil H, Quinn L, van Deursen R, Dawes H, Playle R, Rosser A and Busse M. What effect does a structured home-based exercise programme have on people with Huntington’s Disease? A randomised, controlled pilot study. *Clinical Rehabilitation*. 2013, 27(7): 646 – 658. DOI: 10.1177/0269215512473762

[3.4] Busse M, Quinn, L, DeBono K, Jones K, Collett J, Playle R, Kelly MJ, Simpson SA, Backx K, Wasley D, Dawes H, **Rosser A** and the members of the COMMET-HD management group. A randomised feasibility study of a 12-week community-based exercise programme in people with Huntington's Disease. *Journal of Neurologic Physical Therapy*. 2013, 37 (4): 149-158. DOI: 10.1097/NPT.0000000000000016

[3.5] Busse M, Quinn L, Drew C, Kelson M, Trubey R, McEwan K, Jones C, Townson J, Dawes H, Tudor-Edwards R, **Rosser A**, Hood K. Physical activity self-management and coaching compared to social interaction in Huntington's Disease: Results from the ENGAGE-HD Randomized, Controlled Pilot Feasibility Trial. *Phys Ther*. 2017, 97(6):625-639. DOI: 10.1093/ptj/pzx031

[3.6] Fritz NE, Rao AK, Kegelmeyer D, Kloos A, Busse M, Hartel L, Carrier J, & Quinn L. Physical therapy and exercise interventions in Huntington's Disease: A mixed methods systematic review. *Journal of Huntington's Disease*. 2017, 6(3): 217–235. DOI: 10.3233/JHD-170260

Selected grants:

[G3.1] Busse M & Quinn L. Reliability and minimal detectable change of measures of participation, functional activities and impairments in individuals with Huntington's disease. European Huntington's Disease Network. 1/12/2009 - 1/09/2012; £36,000.

[G3.2] Busse M & Rosser A. Can community supported exercise benefit subjective wellbeing, physical activity levels and abilities in people with Huntington's Disease? Welsh Government HA09/028 06/09/2010- 30/09/2012; £122,720.

[G3.3] Busse-Morris M, Hood K & Rosser A. Supporting activity engagement in people with Huntington's Disease: A phase II evaluation (ENGAGE-HD) Welsh Government Health and Care Research Wales 01/01/2013 - 31/12/2017; £687,952.

4. Details of the impact (indicative maximum 750 words)

Cardiff's research on physiotherapy and physical activity as a positive treatment intervention in HD led to evidence-based national and international guidelines on physiotherapy interventions for HD, as well as training for UK HD health professionals via the Huntington's Disease Association. The research also led to patient resources for physical activity, now being used internationally, including during the COVID-19 lockdown.

4.1 International physiotherapy and multi-disciplinary guidelines for HD

a. The European Physiotherapy Clinical Guidelines

Just prior to the current REF period, the Cardiff team authored the first standardised physiotherapy guidelines for HD, based on the Cardiff research [see 3.1 - 3.5]: the European Physiotherapy Clinical Guidelines, published in English by the European Huntington's Disease Network (EHDN) in June 2013. During this REF period, these were subsequently translated into eight European languages: Danish, Dutch, Finnish, French, German, Italian, Norwegian and Polish, providing HD consultants across Europe with access to evidence-based clinical guidance which they have promoted to other healthcare professionals and patients.

For example, Professor Ed Wild, Associate Director of the UCL Huntington's Disease Centre, noted how he used the guidelines: "*I refer to the guidance in almost every clinic letter I write to GPs...On numerous occasions I have received feedback from patients that the therapists found the [EHDN guidelines] document informative and practically helpful, generally resulting in improved outcomes from the patient's perspective*" [5.1a]. Hanne Ludt Fossmo, physiotherapist at the Vikersund Bad Rehabilitation Centre in Norway, also noted that "*we have used the translated version whenever we are in contact with new patients*", and that "*they are beneficial especially for physiotherapists that have not been in contact with HD, and for newly diagnosed patients*" [5.1b].

b. Influencing HD guidelines worldwide

The European Physiotherapy Clinical Guidelines, and outcomes from Cardiff research, laid the foundation for further inclusion of key recommendations on physiotherapy interventions in significant clinical guidelines for HD management, including:

- i) **British Medical Journal Huntington's Disease Best Practice Guideline** (April 2018) [5.2a], which draws upon Cardiff's home-based exercise research [3.4] and advises that "Home exercise programmes have been shown to improve physical function" [5.2a, p20].
- ii) **The International Guidelines for the Treatment of Huntington's Disease**, were developed between 2015-2018, and published in 2019 [5.2b]. The Guidelines provide global, evidence-based recommendations for everyday clinical practice which standardise all aspects of HD treatment (pharmacological, surgical and non-pharmacological) to improve the patient care and quality of life. The Guidelines list ten motor disorders seen in HD and recommend physiotherapy for five of them based on research from the Cardiff team [3.5].
- iii) **Clinical Recommendations to Guide Physical Therapy Practice for Huntington's Disease** [5.2c], the first global, evidence-based guidelines for physiotherapists to use with people with HD. Published in 2020 in *Neurology*, the Clinical Recommendations cite research papers by the Cardiff team [3.4, 3.5], as well as their European Physiotherapy Clinical Guidelines. The Clinical Recommendations advocate physiotherapy as a key intervention in the management of HD-related dystonia (uncontrollable movement), rigidity, gait and balance disorders, and for assisting with manual dexterity. The Clinical Recommendations were endorsed by the American Academy of Neurology [5.2c, p72] and their publication drew considerable media attention, with podcasts, interviews, and press releases [5.3].
- iv) **Clinical Practice Guidelines on Core Outcome Measures for Adults With Neurologic Impairment Undergoing Rehabilitation** [5.2d] used Cardiff's research to promote the use of outcome measures (e.g., use of the 6 minute Walk Test) [3.3, 3.4]. The Guidelines were published by the American Physical Therapy Association (APTA), an organisation that represents more than 100,000 member physiotherapists, physiotherapist assistants, and students of physiotherapy across the United States.

4.2 Training UK HD health professionals

Following their work on national and international HD guidelines, the Cardiff group collaborated with the Huntington's Disease Association (HDA) of England and Wales to deliver an accredited health professionals' course. This ran from 2015 onwards and has been attended by more than 250 HD health professionals. The Cardiff team used their research findings [3.1, 3.4, 3.5] to produce teaching materials and lead sessions on evidence-based, person-centered approaches to physiotherapy in HD. Course evaluations indicated greater awareness among participants of the positive benefits of physiotherapy and physical activity; feedback from the 2019 HDA course included: "*Good information which I can relay to health professionals in our area to attempt early intervention*" and "*I am better equipped to support them [patients with HD] better with the knowledge I have gained through this course*" [5.4].

4.3 Patient resources for physical activity used internationally

The Cardiff team's patient resources (the *Move to Exercise* DVD [3.4], and the workbook developed through the ENGAGE-HD intervention [3.5]) have been used by patients and clinicians internationally. The resources are available on the HDA's website with 9,906 views by 6,368 individual users between 2019 and 2020 [5.5]. The materials have been translated into Arabic, Italian, Spanish, Portuguese and Chinese, and are being used by health professionals to support patients within these countries. For example, the Chinese materials have been distributed to more than 800 HD families (each with an average of two HD patients) across China to date, with one patient reporting "*I know now that exercise is key to stay healthy*" [5.6]. Xi Cao, President of the Chinese Huntington's Disease Association stated: "*We*

find the exercise video and workbook a strong and practical tool to raise awareness in HD patients and families about importance of exercise and HD patients are using them. Your research into exercise and physiotherapy for management of HD has been very influential to us, and to the advice we give our members” [5.6].

Cardiff's research also led the European Huntington Association (EHA), which supports over 30,000 people with HD, to initiate an exercise initiative, *HD on the Move*, to encourage exercise for people with HD [5.7a]. Based on the research findings [3.4, 3.5], *HD on the Move* encourages and supports HD patients in 28 European countries to be physically active for as long as possible. The initiative is now regularly featured in the programme of the annual EHA family meeting (an annual event bringing together families of HD patients) [5.7b, p8, p12].

4.4 Supporting HD patients during the COVID-19 pandemic

During the first national COVID-19 lockdown, support services for UK HD patients were stopped, with some patients shielding throughout the pandemic. Similar measures were put in place in countries around the world, severely limiting opportunities for exercise. In response, Cardiff's *Move to Exercise* (Section 4.3) was distributed via EHDN mailing lists and licensed for use by HD charity associations in Spain, Italy, Australia, Tasmania, Germany and Brazil [5.8a, b]. The Cardiff team delivered an online exercise session for people with HD in April 2020 with 34 HD attendees from the UK, USA, Netherlands, Norway, Spain and Ireland. 21 attendees rated the webinar as “very helpful”. Feedback included a specialist personal trainer stating: “*I am currently looking at how I can continue working with my HD clients, [after the closure of their facility through government restrictions] and now I can't wait to do remote sessions*” and “*At the moment we go out for a short walk each day, sometimes not far if X is in a lot of pain, so we can try some of the things suggested*” [5.8c]. Available via the UK HDA website, the webinar has received 191 views [5.8d, e].

In summary, Cardiff's research on the benefits of physiotherapy for HD changed international clinical practice guidelines, supported enhanced training for healthcare practitioners, and empowered patients to use exercise and physiotherapy to manage the physical symptoms of a degenerative disease with no other treatments. Astri Arnesen, Chief Executive of the UK Huntington's Disease Association, stated that the improved access to expert physiotherapists and resources for people with HD has been “*very influential in them maintaining their independence for as long as possible – and clearly improves their quality of life*” [5.9].

5. Sources to corroborate the impact (indicative maximum of 10 references)

- [5.1] Testimonials: **a.** Professor Ed Wild, consultant neurologist and Associate Director of the UCL Huntington's Disease Centre **b.** Hanne Ludt Fossmo, physiotherapist, Vikersund Bad Rehabilitation Centre, Norway
- [5.2] **a.** BMJ Best Practice: Huntington's Disease April 2018, **b.** International Guidelines for the treatment of Huntington's Disease 2019, **c.** Clinical recommendations to guide physical therapy practice for Huntington's Disease 2020, **d.** A Core Set of Outcome Measures for Adults with Neurologic Conditions Undergoing Rehabilitation 2018
- [5.3] Podcasts and interviews on the publication of the new international guidance (2020)
- [5.4] HDA Understanding Huntington's Disease, A Certified Course for Professionals. 21-23 May, 2019. Delegate feedback p 4
- [5.5] **a.** HDA website viewing statistics **b.** HDA website material and Handbook
- [5.6] Testimonial: President of the Chinese HD Association
- [5.7] **a.** Testimonial: Astri Arnesen, President of the European Huntington's Association **b.** Programme for the 2019 European Huntington's Association meeting
- [5.8] **a.** EHDN official memo **b.** Move to Exercise covering note and programme workbook (2020) **c.** Summary of feedback from webinar (online exercise session, April 2020) **d.** UK HDA website featuring webinar **e.** Screenshot of webinar recording on Vimeo
- [5.9] Testimonial: HDA Chief Executive