A Study of What Helps People with Huntington's Disease Live with their Condition.

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Abstract

Background
Huntington's disease (HD) is a life-limiting neurodegenerative condition in which the onset of symptoms, generally in midlife, is characterised by cognitive decline, psychological problems and movement disorder. HD is inherited as a dominant genetic trait. Co-morbities are prevalent and include psychiatric effects such as depression, anxiety and psychosis. Death from suicide among people with HD is reported to occur four to five times more frequently than among the general population. The only treatment for HD supported by well established evidence for effectiveness is tetrabenazine for reducing chorea and one of its side effects is depression.

Aim
To identify and prioritise "what helps people with Huntington's disease live with their condition" from the perspectives of people with Huntington's disease, their carers, and healthcare professionals who treat them

Method
People with HD, family caregivers and health professionals generated 94 statements in answer to the question: 'what helps people with HD live with their condition?' The participants were based in community and institutional contexts. They prioritised the statements by organising them into five levels of importance, least to most important. Finally the participants arranged the statements into groups or 'clusters' to indicate which statements seemed subjectively to each participant to 'belong' together. Specialised 'concept mapping' software (Ariadne ®) was used to graphically depict the consensus among participants of how the statements were clustered and prioritised.

Results
Expert assessment and treatment for co-morbid mental health problems were the highest priority for all participants. Flexibility in the provision of care and timeliness of help provided through integrated interdisciplinary teamworking with specialist HD expertise were also high priorities. People with HD prioritised being trusted and supported to maintain independence. They placed greater value on medical treatments than caregivers or professionals, who were more concerned with care and planning for future needs in more advanced stages of the disease. Overall participants were more concerned with lessening the impact of symptoms on living with HD than with measurably reducing specific symptoms such as chorea.

Conclusions
Healthcare professionals can help people with HD live with their condition by being more proactive in identifying and treating mental health problems, which is at present an important unmet need.

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Glossary of Abbreviations

AD	Alzheimer's disease
DoH	Department of Health (UK)
DLB	Dementia with Lewy Bodies
EHDN	European Huntington's Disease Network
HCP	Health Care Professionals
HD	Huntington's disease
HDA	Huntington's Disease Association
MS	Multiple sclerosis
NHS	National Health Service
ONS	Office for National Statistics
PD	Parkinson's disease
PwHD	People with HD
SHA	Scottish Huntington's Association
TBZ	Tetrabenazine

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Chapter One
Introduction

Due to the complexity of its effects on individuals and families the treatment, care and service provision for people affected by Huntington's disease (HD) poses a difficult challenge to professionals (Aubeeluck and Wilson 2008). As the disease progresses often over many years, the symptoms negatively impact on every area of independent living (Lechich et al 2008). There is reportedly no cure for HD or indeed any effective treatment aimed at slowing the deterioration of the brain as a result of the condition (Ramaswamy et al 2007; Adam and Jankovic 2008; van der Burg 2009) and the absence of curative therapies adds to the importance of the question that this thesis seeks to answer: what helps people to live with HD?

This study considers the viewpoints of people who have HD, their caregivers and healthcare professionals who treat them, about what helps people with HD to live with the condition. Knowledge of these perspectives is needed to ensure that professionals who provide care and treatment and who decide on the focus areas for HD research are informed about what matters most to those affected by the disorder, and any differences between the perspectives that should be taken into account.

1.1 What is Huntington's Disease?

Huntington's disease (HD) is a neurodegenerative disorder that commonly manifests between the ages of 35-50 (Handley et al 2006). Symptoms can be divided into three groups- motor impairment, psychiatric problems and cognitive deterioration (Langbehn et al 2004) and the mean length of time between onset of symptoms and death is reported to be in the region of 15-20 years (Handley et al 2006). The condition is inherited as a dominant genetic trait (Bonelli and Hofmann 2007) meaning that offspring are at 50% risk of developing the condition at some time in

their life. The responsible mutation was identified on the short arm of chromosome 4 in 1993 (Gusella et al 1993; Huntington's Disease Collaborative Research Group 1993) making genetic testing for HD possible.

1.2 Pathophysiology

The HD gene 'IT15' is necessary for life and is responsible for regulating the protein 'huntingtin'. This protein is important for brain function and less widely appreciated is its expression more generally in body tissues and organs (Li et al 1993). IT15 consists of a repetition of three nucleotide bases- cytosine, adenine and guanine (or CAG). The normal range for the number of CAG repeats in the gene has been reported to be between 13 and 31 (Sanchez et al 1997) but in HD it could be between 36 and 250 although many individuals with up to 40 repeats do not develop HD (Langbehn et al 2004). These authors demonstrated a negative correlation between CAG repeat length and age of onset meaning that higher repeat number is associated with manifestation of the disease in younger people. More rapid brain deterioration following diagnosis and greater severity of symptoms are also associated with a greater CAG repeat number (Ruocco et al 2008).

This trinucleotide expansion disrupts control of 'huntingtin' (Caviston and Holzbar 2009). Through a process that has yet to be established this causes degeneration of neurones within the brain- specifically the basal nuclei and cerebral cortex (van der Burg et al 2009)- accounting for the movement disorder and cognitive and neuropsychiatric problems discussed below. However, van der Burg et al (2009) state that HD is not only a brain disorder as it also has 'peripheral features' including weight loss, skeletal-muscle atrophy, cardiac failure, testicular atrophy and osteoporosis. Within the brain itself appreciation for wider deterioration than initially

envisaged has grown in the last two decades before which time HD was characterised by the loss of striatal neurons (Reiner et al 1988). More recently the likelihood that clinical deficits relate to deterioration of the cortico-basal nuclei network rather than to damage restricted to the striatum is appreciated (Delmaire et al 2010).

1.3 Detection of disease onset and clinical diagnosis

Modern imaging techniques can detect structural and functional changes in the brain prior to the observable onset of symptoms (Squitieri et al 2009, Klöppel et al 2009). Paulsen et al (2007) demonstrated that measurable disease development can occur ten to twenty years before diagnosis as physical, cognitive and psychological impairment is subtle initially and progresses gradually (Harper 2005). There is controversy concerning which of these areas is the site of the first changes and a lack of uniformity in presentation between individuals appears to explain the disparity within the literature (Snowdon et al 2002). According to the UK NHS (National Health Service (NHS) 2013) behavioural changes are frequently the first apparent signs in a person who is developing HD but many individuals refuse to accept there is any problem at this stage. Clinical diagnosis is made on the basis of observed motor signs occurring in an individual with a family history of HD and this diagnosis is regarded as confirmed by a positive test for the expanded HD gene (Witjes-Ané et al 2007). It follows that people newly-diagnosed with HD may have relatives, friends and colleagues who become aware of problems long before this confirmation.

1.4 Effects of Huntington's disease

Cognitive decline

Neuronal degeneration in HD is selective (Li et al 2001) and initially the striatum (part of the basal nuclei including the putamen and caudate nucleus) are affected and eventually damage becomes more widespread, particularly involving the frontal and temporal cortex (Montoya et al 2006). Disruption to these areas is associated with impaired attention, working memory and executive functions. Executive functions are necessary for ability to divide or switch attention between competing tasks and are thought of as supervisory cognitive processes in managing the brain's cognitive skills (Alvarez and Emory 2006).

Psychomotor speed - the relationship between mental processes and movement or muscular activity - is impaired as are language functions (Podoll et al 1988; Teichmann et al 2008), problem-solving ability and visuo-perception (Salmon et al 2001; Salmon and Heindel 1998). Planning and organising daily activities become problematic and thinking is increasingly rigid and inflexible so that the person is easily 'stuck' continuously rehearsing a thought or a task (Aubeeluck and Wilson 2008) and it becomes difficult to accept changes to routine (Pollard 2008). Problems with speech and language result from motor disorder and also from difficulty in organising retrieval of information from semantic memory. People with HD tend not to initiate conversation and prefer to use short phrases or single words when spoken to and there may be long silences prior to a response to a question (Aubeeluck and Wilson 2008; Pollard 2008).

Memory function is impaired but this occurs later in the course of the disease process and is different to memory loss in dementia such as in Alzheimer's disease (Peavy et

al 2010). Loss of memory function in HD is likely to be secondary to other cognitive deficits whereas in Azleheimer's type dementia, memory loss features more prominently as a primary disease effect (Peavy et al 2010). Apathy and impulsive behaviour (disinhibition) can be detected in people who are positive for the HD genetic mutation prior to diagnosis and these initially subtle effects increase in relation to progression of motor disorder (Duff et al 2010) .

Motor impairment.

Typically the first visually noticeable signs of HD are fidgeting, twitching fingers and general restlessness (McNeil 2009). Involuntary jerking 'choreic' limb movements develop over time and eventually the trunk may writhe and twist. Although chorea is widely known as a characteristic feature of HD (Ramaswamy et al 2007), Phillips et al (2008) suggest that many patients do not seem to find it unduly problematic. Other associated movement disorders, commonly rigidity, dystonia and bradykinesia tend to become more prominent symptoms as chorea diminishes over time and are usually more disabling (Nance et al 2003).

Despite the reports that many do not find chorea in HD greatly inconvenient Nance et al (2003) suggest that although many patients with HD are unaware of it, it should be acknowledged that some find it not only physically but also socially disabling (Oates et al 2006). It generally persists through all waking hours only subsiding during sleep and is aggravated by stress and anxiety (Rosenblatt et al 1999[a&b]).

Speech deteriorates with cognitive decline, aggravating the problems of dysarthria (Nance et al 2003). Breathing is increasingly problematic as impaired motor control affects intercostal muscles and the diaphragm. Swallowing also becomes problematic

and in later stages of HD the most common cause of death is dysphagia leading to choking, aspiration or malnutrition (Nance et al 2003).

Psychiatric problems

Psychiatric disturbance is reported to occur frequently prior to the manifestation of motor or cognitive disorder in HD (Julien et al 2007). These authors found that depression presenting at this stage cannot be accounted for only by the natural concerns associated with living at risk of developing HD. Depression, anxiety, apathy, compulsive behaviour and psychotic episodes are among the conditions associated with HD (van Duijin et al 2007).

In the past apathy has often been referred to as a psychiatric symptom in HD (Folstein and Folstein 1983). Thompson et al (2012) refer to apathy, irritability and depression associated with HD as neuropsychiatric symptoms. While their recent study involving neuropsychiatric assessments of 111 people with HD found that irritability increased only in early stages of the condition and depression occurring with HD did not increase at any stage, they found that apathy increased with the progression of HD and appears 'intrinsically linked' to the disease trajectory. Thompson et al (2012) concluded that previous studies may have underestimated neuropsychiatric symptoms as a problem occurring with HD.

The risk of suicide in HD is greater than that among the general population (Novak and Tabrizi 2010). Paulsen et al (2005[a]) found that suicidal ideation among the participants with the HD genetic mutation was experienced most frequently at the time when the person was near to the threshold of being clinically diagnosed with HD. Depression and impulsivity are reported as risk factors associated with suicide in HD (Craufurd and Snowden 2002). However, Paulsen et al (2005[a]) suggest that

suicidal thoughts in HD can occur without depression and can sometimes be considered a rational response to having the condition.

Stigma

Difficulty in determining the prevalence of HD is attributed by Rawlins (2010) to a tendency within many families to avoid acknowledging symptoms or the presence of HD due to the stigma associated with the disease, which led to significantly lower estimates of prevalence in the past than likely to be true. In support of this proposed explanation, Rawlins (2010) cited a graphic historical account by Wexler (2010) which reports a number of specific examples of social stigma experienced by families affected by HD.

1.5 Epidemiology

Prevalence

Accurate and up-to-date figures of the prevalence of Huntington's disease from reliable sources do not appear to be available. Harper (1992) calculated estimates using population surveys and analysis of death rates and concluded that the prevalence of HD in North and South European countries is somewhere between four and eight per 100,000 population. Harper (1992) also noted differences internationally and suggested a similarly high occurrence in some Asian regions and in India but lower prevalence in Finland and Japan.

Based on a UK population of over 63,000,000 (Office of National Statistics (ONS) 2013) Harper's (1992) European estimate would suggest that there are likely to be between 2,500 and 5000 people in the United Kingdom (UK) with a confirmed HD diagnosis. In 2002 Harper narrowed this estimate of prevalence in the UK to 6-7 per

100,000 population (Harper 2002) and this would equate to between 3600 and 4200 in a 63,000,000 UK population. More recently Rawlins (2010) challenged this and basing his calculations on numbers registered with the Huntington's Disease Association and claimed that the UK prevalence might well be in excess of 12.4 per 100,000. Hoppitt et al (2010) accessed a primary care database holding data on seven million patients registered at 479 GP practices and calculated a prevalence of HD of between 5.96 and 6.54 per 100,000. They suggested that if patients with unspecified chorea and patients undiagnosed with a family history of HD were taken into account a prevalence of 10 per 100,000 population seemed a reasonable maximum estimate. Hoppitt et al (2010) stated that the higher figure proposed by Rawlins (2010) needed further support.

Loy et al (2010) report that their experience in clinics in New South Wales fits with Rawlins's suggestion of increasing HD prevalence in the UK and they offer additional reasons to that put forward by Rawlins. They state that 'the baby boomers' are now in their 50's and 60's; the age of peak HD prevalence. They also state that they continue to encounter new patients with no family history of HD and cite the claim by Ramos-Arroyo et al (2005) that 4% of HD cases are due to new mutations (i.e. are not inherited).

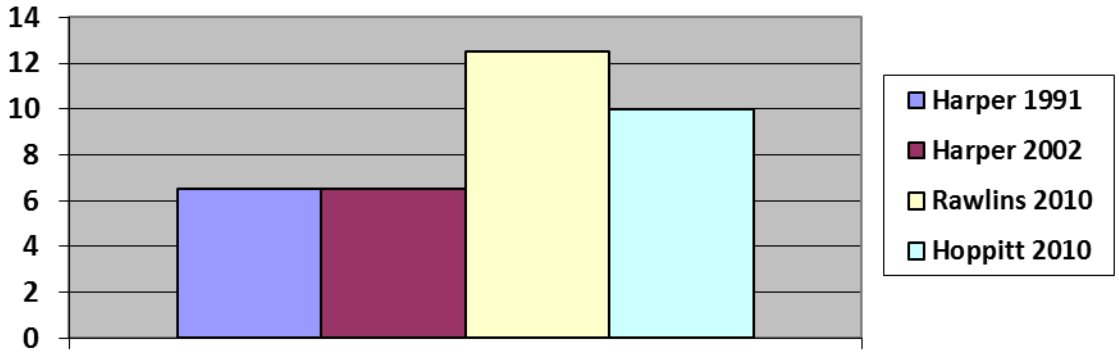


Fig 1.1 Estimated HD prevalence per 100,000 population. The middle estimate (Hoppitt et al 2010) would equate with 6,000 people diagnosed with HD in Britain.

Fig 1.1 presents a comparison of estimates of the prevalence of HD with Hoppit et al's (2010) figure of 10 per 100,000 population appearing to mediate between extremes, arguably making suggestions of 6,000 people in Britain diagnosed with HD realistic. However, the debate seems set to continue, with Sackley et al (2011) concluding that while evidence to support Rawlins' estimate may be lacking, the true figure may well be much higher than has been demonstrated thus far.

Movement disorder

According to Kremer (2002) chorea occurs in 90% of cases of HD and Louis et al (1999) cited two neurologists who reviewed video-tapes of 42 people with HD and found that 95% had dystonia of differing severity. For seven (16.7%) of these individuals, the dystonia was found to be severe and constant while 24 (nearly 60%) were troubled by this in at least one body area more than half of the time. A later paper involving 127 patients suggested that for around 12% of the same subjects, dystonia was the predominant HD symptom (Louis et al 2000).

Fig 1.2 shows that while chorea is the most noted symptom of HD (Ramaswamy et al 2007), dystonia is slightly more prevalent in people with HD and is more physically disabling (Louis et al 2000).

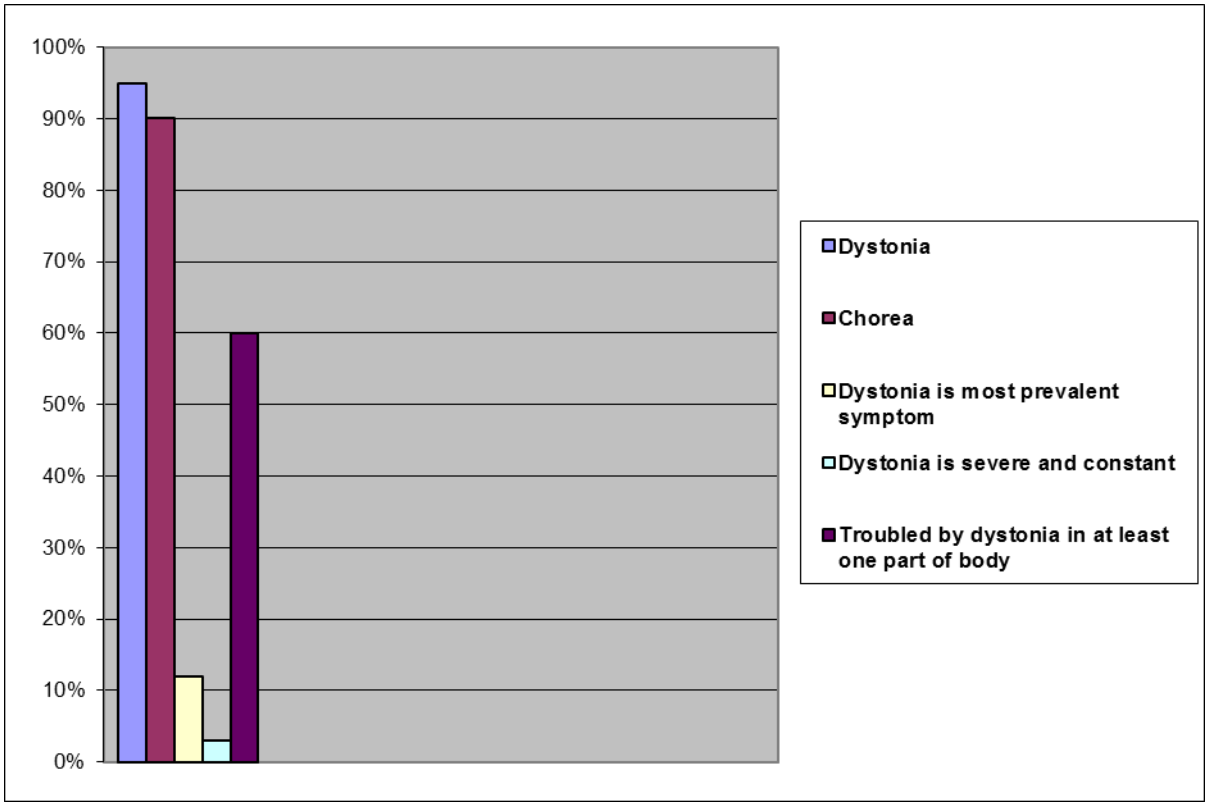


Fig 1.2 Prevalence of movement disorder in people with HD by type, severity and predominance, based on studies cited above.

Psychiatric problems

In a survey of over 4000 carriers of the HD genetic mutation 17.5% reported experiencing suicidal thoughts at the time of the survey and 10% had made at least one suicide attempt previously (Paulsen et al 2005[b]). It is difficult to state the proportion of people with confirmed HD affected by psychiatric problems with any

certainty. Estimates range from between around one third (Cummings 1995) to three quarters (Watt and Seller 1993). These estimates are depicted in Fig 1.3.

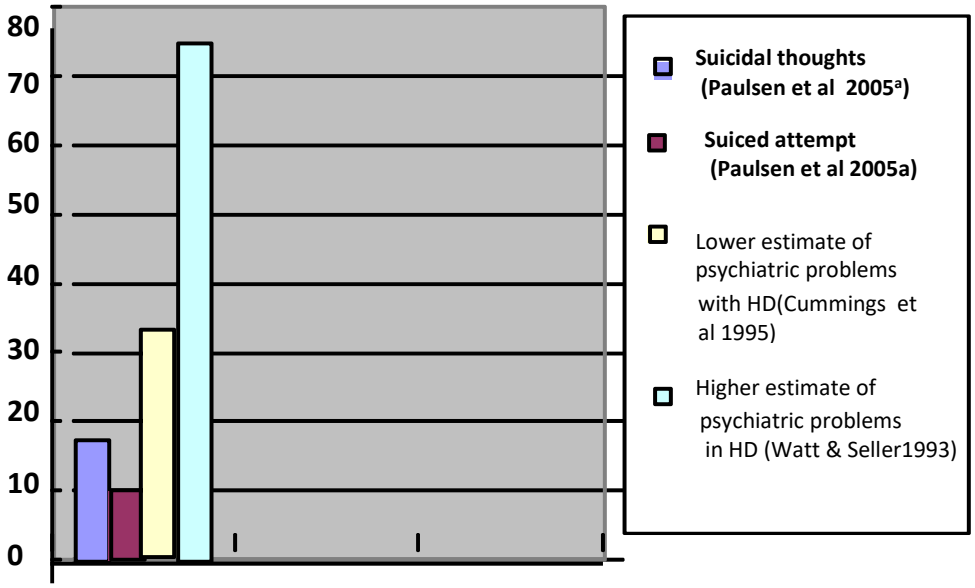


Fig 1.3 percentages of psychiatric problems and suicide ideas and attempts among people with HD according to cited sources above

1.6 Genetic Inheritance

The dominant pattern of inheritance in HD means that a person with one mutated HD gene will develop HD at some time in life providing she or he does not die earlier of some other cause. This is the case despite having one normal copy of the gene in nearly all cases. Newly conceived offspring inherit one allele (form of the gene) from each parent. Assuming that a partner of a person with this HD mutation has two normal alleles, a child of this couple will definitely possess one normal allele coming from the mutation-free parent. From the affected parent, the child may have inherited the normal copy or the mutated copy, hence the 50% risk.

HD is described as a single-gene disorder (e.g. Chial 2008; Hormozian et al 2004) yet modifying factors are known to play a part in determining the age of onset and

the pathogenic process (Swami et al 2009). People with between 27-35 CAG repeats on the HD gene (IT15) are said to be within a 'high normal' range (Hendricks et al 2009) and no-one within that range has had a confirmed diagnosis of HD. Yet some fathers but not mothers within that range may pass on a penetrant HD genetic mutation (more than 40 CAG repeats) to offspring and this means that anyone with more than 40 repeats will develop HD at some time in life unless they die of some other cause before onset. That is, the expanded gene is fully (100%) penetrant. It should be added that despite the established association between higher numbers of CAG repeats and earlier HD onset this is merely a trend. Age of onset cannot be predicted on the basis of the length of an individual's CAG count (Squtieri et al 2009). It seems fair to conclude that a description of HD as a single gene disorder may understate the complexity of factors associated with inheritance.

Predictive testing for HD has been available since 1987 following the identification of a genetic marker on the short arm of chromosome four (Creighton et al 2003). The linkage test depended on samples from various family members to maximise the likelihood of up to 95% accuracy. The Hereditary Disease Foundation (1994) reported that many people who wished to be tested experienced difficulty in recruiting family members to take part leading to lower confidence in test results or to the procedure becoming a non-feasible option. The HD genetic mutation was identified in March 1993, a decade after the genetic marker leading to the development of a near to 100% accurate genetic test available in the UK from late 1993 (Harper et al 2000). Most tests in the UK are provided with formal structured counselling based on international guidelines (Tibben 2006) at Clinical Genetics Departments within the National Health Service. The test can inform people who are at risk of HD whether they have the mutation or not. For people who have symptoms

akin to those associated with HD the test can rule out the condition or provide confirmation of the diagnosis. Prenatal testing can also determine whether an unborn child has the genetic mutation for HD (Wolff 1996).

Decisions about whether to be tested for HD are complex and some of the reasons for agreeing to the testing for HD include the wish to reduce anxiety associated with uncertainty concerning risk status and to aid decisions about family planning. Reasons for turning down the offer of testing for HD might include fear concerning loss of health insurance or mortgage opportunities or of confirmation of the presence of the mutated gene and its future consequences. Due to the complexity surrounding the choice, family members tend to feel strongly about whether or not individuals should be tested and often disagree (Klitzman et al 2007).

1.7 Impact on individuals and families

Families affected by HD are faced with many challenging dilemmas. For example they may have to make decisions such as: who will care for sick family members; how and when to tell children about their HD risk status; whether or not to have a genetic test; is it useful to be prescribed a particular medication; whether to participate in a research study; whether to have a tube inserted into the stomach when it is no longer possible to swallow safely; or to donate one's brain for research or education. Choices may place a heavy sense of responsibility onto people already struggling to cope with daily activities.

People diagnosed with HD could face losses such as job prospects and may experience loss of control in the form of other people having to manage their finances, driving, etc. Family members may also have to manage unusual or

sometimes aggressive behaviour from the person with HD who may depend on their assistance to get to the toilet, have a bath, eat or dress.

The complexity of these aspects of the impact of HD on the diagnosed person and family members, friends and caregivers are outlined in numerous texts (e.g. Aubeeluck and Wilson 2008; Adam and Jankovic, 2008; and Lechich et al 2008). The diagnostic label 'Huntington's disease' cannot adequately capture this complexity and the particular ways that different individuals and families experience the impact of the condition as is true of any other medical diagnostic label.

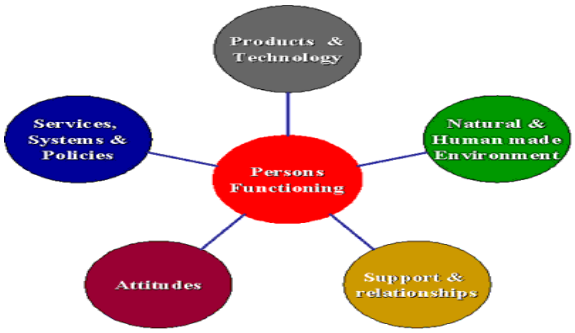
Recognising this problem, the World Health Organisation (WHO 2001) promotes consideration of the impact of disability on individuals and their families in terms of the International Classification of Functioning (ICF). This framework encourages health professionals to take into account the impact of the health condition on body functioning and body structures, activities and social participation. Although the model was developed for the purpose of classifying conditions it is helpful here towards contemplating the broad impact of HD on individuals and families as HD presents families with complex problems in all of the depicted areas.

Fig 1.4a illustrates factors that can contribute to a person's functioning. Tomey and Sowers (2009) highlight the inadequacy of attempting to appreciate the impact of physical functioning without context. Environmental, social and service provision factors tend to be adapted to compensate when functioning is compromised (Tomey and Sowers 2009). For example medication, handrails, a hearing aid or adapted transport services may prevent a personal function limitation becoming disabling.

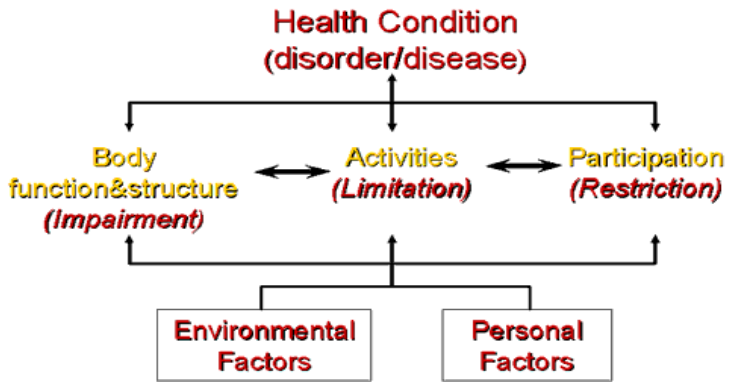
Figure 1.4b demonstrates the interactive relationship between the health condition (HD) and environmental and personal factors on body function and structure, activities and participation (as explained in the introduction).

Fig 1.4 A &B: The WHO ICF Framework (WHO 2001).

A:



B:



Source: WHO, http://www.who.int/classifications/icf/en/ P18, *Reproduced with permission.*

The method selected in this thesis to examine what helps people with HD live with their condition needs to be capable of facilitating all of ICF components. The justification (in chapter two) for choosing a method known as 'concept mapping' will demonstrate that this is an inclusive rather than consensus design. The concept mapping method engages participants (who in this case will be people with HD,

caregivers and health professionals) in generating statements intended to convey factors that are helpful in living with HD. Rather than seek a consensus about what factors should be considered concept mapping will ensure that any factor considered relevant even by one participant will be represented. In the discussion section at the end of this thesis the extent to which the method has enabled people to raise a sufficiently broad range of factors will be considered. The ICF is intended as a framework capable of capturing the whole impact of a health condition and so a successful concept map would be expected to contain some items (statements) relating to each of the components of the model.

1.8 Treatment

While a range of available interventions are prescribed to improve symptoms associated with HD, a systematic review designed to establish whether any pharmaceutical treatments have been shown to have consistent control of HD symptoms concluded that only one medication was shown to be effective (Mestre et al 2009[a]). The drug 'Tetrabenazine' showed clear efficacy for the control of chorea, but was reported to be associated with serious adverse effects. Clear effectiveness was not demonstrated in trials of any other medications for HD and Mestre et al (2009[a]) called for well-designed randomised controlled trials in this area.

In another systematic review, Mestre et al (2009[b]) found that no pharmacological treatment was effective as a disease-modifying therapy for HD. Mestre and Ferriera (2012) undertook a further review using a systematic approach and found that while it is encouraging that many newly developed interventions have been the subject of recent and current clinical trials, Tetrabenazine remains the only treatment for HD (for chorea only) with efficacy supported by strong clinical evidence. Mestre and

Ferriera (2012) suggest that the clinical management of HD needs to rely on recommended good practice based on low level evidence and on efficacy demonstrated in trials of therapeutic interventions for other disorders in which people who are affected may experience similar symptoms and disease impact.

Experiences of living with HD and with other similar disorders are considered below as are the responses to specific problems that have been made by health care professionals, caregivers and people directly affected by HD. This is intended to provide context for the research question that this thesis will investigate.

This thesis seeks to add to knowledge and understanding of perspectives of those directly affected by HD and their family caregivers and of health professionals with experience of caring for people with HD, about what helps people to live with the condition. In the absence of a cure it is important to work to assist those who provide appropriate treatment, care and services and those who decide the focus for research examining effectiveness of interventions for HD to do so with an appreciation of priorities from these perspectives. The purpose of the thesis is to contribute to such understanding.

To this end the next chapter examines experiences of people with HD and other similar conditions and responses to these experiences. A rationale for the choice of study method- concept mapping- aimed at establishing perspectives of what helps people live with HD is provided. Chapter Three explains the methodology, Chapter Four presents results and the final chapter discusses the implications of the findings.

Chapter Two
Literature review and rationale for the study

This review of the literature aims to describe the patient experience of Huntington's disease (HD) and also the responses of patients, carers and healthcare professionals to these challenges. Evidence concerning the subjective experience of living with HD is limited and so it is necessary to consider studies that have investigated patient perspectives of living with other diseases that share some features with HD such as movement disorders, mental health problems or deteriorating cognition. The literature shows that problems identified in studies concerned with subjective experiences of people with HD and their family caregivers also feature in investigations of experiences of people with other degenerative neurological conditions such as Alzheimer's disease (AD) and other forms of dementia, Parkinson's disease (PD) and multiple sclerosis (MS). Some experiences of people who live with impairment resulting from stroke may also be relevant. In the context of movement disorder for example, Quinn et al (2013) suggest that while the symptoms of Parkinson's disease and Huntington's disease are unique the two conditions do share aspects that have relevance to rehabilitation.

The review was informed by a search of literature obtained from AMED, CINAHL, MEDLINE, EMBASE, PsychINFO and Cochrane Library databases. Each database was searched from inception using the keywords: Huntington's Disease; Neurological; Parkinson's; Alzheimer's; Multiple Sclerosis; Stroke; Dementia; Care; Carer; Caregiver; Impact; Quality of Life; Experience; Intervention; Treatment. Google Scholar® and Google® were also searched, as were the 'Huntington's Disease Association' and 'European Huntington's Disease Network' websites. Presentations and informal discussion with experts in the field of research in

interventions for HD at the following conference proceedings were also part of the search strategy:

- The European Huntington's Disease Network 6th EHDN Plenary Meeting: Roadmap for Effective Therapies in Huntington's Disease, Prague, September 2010.
- Movement Disorders Symposium, Lund University, Sweden, March 2011.
- Movement Disorders Society 16th International Congress, Dublin June 2012.

Papers were examined if the focus was considerd to be concerned with the experience of living with a neurological disorder or with patient, caregiver or health professional perspectives about what helps people live with these conditions.

2.1 Responses to experiences of HD diagnosis and genetic testing.

The abnormality that constitutes the genetic basis for HD is an excessive number of repeats of the trinucleotides CAG on the short arm of chromosome four. However despite the knowledge that more than 40 repetitions cause HD (Novak and Tabrizi 2010) and more rapid progression is associated with higher repeat numbers, the date of onset and the rate of deterioration for individuals cannot be predicted (Paulsen 2011).

Erwin et al (2010) reported a high level of perceived stigma and discrimination associated with a family history of or genetic testing for HD (46% of 433 participants in the USA, Canada, Australia) regarding insurance, employment and relationships. Anxiety about discrimination and stigma resulting from a diagnosis of HD was reported to be a greater problem than the actual negative experiences. The extent to which this may deter people from seeking a diagnosis is not known (Erwin

et al 2010). Asscher and Koops (2010) defend ‘the right not to know about Huntington’s’ in response to a Dutch government decision to prevent couples from having preimplantation genetic diagnosis (PGD) without the at-risk parent knowing their own HD risk status. Medically it is perfectly possible to select an embryo without sharing the diagnosis (whether positive or negative) with the at-risk parent. Asscher and Koops (2010) suggest that the importance of the right not to have the burdensome knowledge is greatly underestimated. However Novak and Tabrizi (2010) report that a HD positive diagnosis can be a relief for some people as this removes uncertainty which can be harder to live with than coming to terms with the reality.

Currently diagnosis is founded on neurological assessment confirming associated movement disorder in conjunction with a positive genetic test for the HD mutation (Paulsen 2011). A positive genetic test in the absence of clinically identifiable motor signs indicates that the person is in what is termed a prodromal phase (Paulsen 2011: p2). This situation is felt to be unsatisfactory given that the presence of detrimental cognitive effects has been established as evident at least 15 years prior to detection of motor symptoms (Paulsen et al 2006, Paulsen et al 2007). Revision of the diagnostic criteria involving reliable and valid cognitive measures is advocated (Paulsen 2011).

A number of studies focussed on fear and uncertainty related to HD (e.g. Semaka et al, 2012; Huniche, 2009; Dufrasne et al, 2011), and this tends to be associated with the hereditary nature of the disorder and to problems related to decisions regarding whether to undergo predictive testing for HD. Eddy et al (2011) found evidence of fear and uncertainty specifically relating to predictive or confirmatory testing.

While access to knowledge of genetic status regarding HD can be considered a right for at-risk individuals so can the choice not to know (Taylor 2004) and it is argued that benefits in terms of relief from uncertainty and informed life options and planning override potential disadvantages (Codori and Brandt 1994). From analyses of records to date, while genetic testing raises a variety of practical, social and ethical problems requiring skilful tact and experience from health and social professionals to work with families and individuals to overcome, there is no evidence that providing the information has resulted in harm and none to support the view that receipt of certain results leads to depression (Broadstock et al 2000, Meiser and Dunn 2000, Nanetti et al 2012). This is supported by studies using qualitative interview and also self- report questionnaire approaches (e.g. Pakenham et al 2004, Rouwer-Dudokdewpith et al 2004).

Problems with adjusting to the predictive test results and a person's psychological wellbeing appear from this body of evidence to relate to emotional state prior to being tested and not to the result received.

That decision-making is complex (as is the impact of genetic testing on individuals, couples and families) seems well established but the evidence for predictive factors associated with responses to receiving test results is not strong. There is a need for further good quality research in the areas identified above on which to base continued revision of international protocol, and the need for counsellors to be well trained and experienced, flexible and aware of the complexities raised in the accumulated body of evidence so far is called for (Cordier et al 2012). Meanwhile new guidance for predictive testing for HD concurs with the view that most individuals who undergo the process do not experience negative psychological consequences but emphasise the importance of identifying those who may be

vulnerable and ensuring adequate counselling is offered and provided (MacLeod et al 2013).

2.2 Responses to experiences of fear and uncertainty regarding disease progression

Eddy et al (2011) reported that the experience of fear in people with HD is reduced as the disease progresses. They suggest that changes to fear responses may explain increased risk-taking behaviour with potential detrimental personal and social consequences. However the method involved subjective ratings of emotional responses to pictures and scenarios and as the authors acknowledge, ability to appraise pictures and scenarios may have been a confounding factor. Rating fear responses to these stimuli may be very different to fear experienced as a result of living with a condition with uncertain onset and rate of progression. This literature review has found no recent evidence of fear and uncertainty associated with living with HD other than that specifically relating to predictive or confirmatory testing but that is not to say these emotions are not part of the experience of living with the condition. Wexler (1979) describes fears concerning HD symptoms among at-risk interviewees and reports a particular dread of losing continence and the 'horror' of anticipated body changes. Wexler summarises the interviews in terms of 'fears, griefs and hopes of persons coping with an unusual life situation' (Wexler 1979: pp199-200). Since that time, the problem of uncertainty and fear in HD seems un-investigated but in other degenerative neurological conditions the aspect has been researched (Cole and Vaughan 2005, Steeman et al 2007), and there is arguably no reason to doubt the relevance of findings across diagnoses.

Fear of consequences of changes due to disease manifestation and difficulty living with uncertainty are reported to be common experience in PD (Cole and Vaughan, 2005) and in dementia (Steeman et al 2007). Fear of symptom exacerbation and deterioration and uncertainty regarding the future are also major concerns among people with multiple sclerosis (MS) (McNulty et al 2004).

It seems reasonable to assume that similar experiences of fear and uncertainty are problematic for people with HD and currently there appears to be a lack of literature evaluating any response to this by healthcare professionals, by informal family caregivers or indeed by people with HD themselves, and hence this may be an important area for future exploration.

2.3 Responses to experiences of loss associated with neurological disease

In a recent qualitative study people with HD and family members expressed impaired communication in terms of a loss, which was perceived to negatively impact on meaningful social participation (Hartelius et al 2010). Other losses experienced by families affected by HD have been expressed as loss of personality, loss of physical aptitude in affected parents and loss of the usual family structure (Smolina 2007). People with PD have also been shown to perceive the impact of the condition in terms of losses (Charlton and Barrow 2002). In a qualitative study, eight individuals expressed their experience of the impact of having PD in terms of the loss of their own personal identity and the loss of the ability to function socially, physically and mentally (Charlton and Barrow 2002). Similarly, 'loss of self' or 'personal identity' is commonly expressed among people with Alzheimer's disease (AD) (McRae 2011). Seeing the impact of disease as a series of losses was a core finding of a recent review of 43 qualitative studies exploring subjective experiences

of living with dementia (Steeman et al 2007). These losses were said to threaten perceived security, autonomy and of being a useful or meaningful society member. Fear of further losses in future, reducing the ability to retain the personal identity, or sense of self, were common across the diagnoses in these studies (Charlton and Barrow 2002; McRae 2011; Steeman et al 2007).

A large body of research concerned with older people living alone suggests that chronic disease is often experienced in terms of similar losses to those expressed above, indicating that the problem is not peculiar to neurological disease but is more generic (Haslbeck et al 2012). This systematic review found that it is widely held that these perceived losses can be best compensated for by maintaining control and independence and that support towards self-help and self–management can play an important role in achieving this (Haslbeck et al 2012).

The view seems to be shared in the context of neurological disorders and specifically HD. Helder et al (2002) found illness perceptions and coping mechanisms adopted by people with HD to be predictive of wellbeing in HD. De Boer et al (2007) suggested that their research showed that understanding of the way people with dementia cope should be encouraged among professionals and service providers as the knowledge can help to tailor services and care more appropriately. De Boer et al (2007) found that contrary to widely held perceptions people with dementia passively deteriorate and are helpless to influence the impact of the disease, coping mechanisms are used to confront the effects. De Boer et al (2007) recognised emotion-focussed and problem-focussed (considered more effective) mechanisms being employed and concluded that the finding emphasised the value of teaching effective coping strategies. Given the similar conclusions of Helder et al (2002) concerning HD the importance of coping mechanisms in living with neurological

decline is arguably not disease-specific as far as implications for service and care provision are concerned.

People with HD, their caregivers and health professionals have reported benefits of involvement in self-help support groups in coping with living with the disorder (Dawson et al 2004). Self-help group membership, attendance and participation are considered a factor related to how well people with neurological disorders cope with and adjust to living with the condition. With regard to PD, Charlton and Barrow (2002) found that self-help group attendees used different coping strategies to those not involved in groups, with group members tending to incorporate PD into their lives, embracing the condition, while non-attendees were more likely to cope by striving for a 'normal life'. Ways of coping with PD included adopting a positive attitude, maintaining a fighting spirit, avoiding thinking or talking about PD, having hope, accepting the disease, and keeping up active social engagement (Charlton and Barrow 2002). For non-group attendees, equipment designed to offer practical help to overcome physical problems were more likely to be perceived negatively as a landmark in their deterioration. Schwartz (1999) found that attending a self-help group improved external locus of control for people with MS, although this did not improve psychosocial role performance or wellbeing. Those participants in the study with affective symptoms of MS benefitted most from peer group support. However, Schwartz (1999) concluded that teaching coping skills was more effective than peer group involvement.

Authors of a pilot study examining the feasibility of a patient education programme for people with HD based on a programme developed for people with PD reported that the self-management approach inherent in the programme can improve quality of life in both conditions (Campo et al, 2012). The concept of self-management

which involves providing support for people with long-term health conditions to manage their symptoms and treatment and the physical, social and psychological impact of chronic disease (Department of Health (DoH) 2005[a]) has become central to health policy not only in the UK but also internationally (Coster and Norman 2009). However, Battersby et al (2010) suggest that while the need to empower people with chronic conditions is undisputed, clear information to help professionals to decide specifically how to improve patients' abilities to self-manage is lacking.

Schneider and Young (2010) concluded that professionals should see their role as helping people with MS towards self-management and avoid a tendency to 'take over'. Jones et al (2013) emphasise the importance of self-management in the long-term care of people post-stroke, and identify a lack of research in this area as problematic. Jones et al (2012) report that despite increasing research activity focussed on self-management programmes in stroke care many aspects remain unexplored and they call for collaboration in this area across chronic diagnoses. De Boer et al (2007) also emphasise the importance of working to encourage and facilitate self-management as opposed to giving care that amounts to 'taking over', in their a review of 50 papers concerned with the experiences of people with dementia.

Building on evidence supporting enhanced self-management through home-based exercise programmes in stroke (Olney et al 2006) and PD (Lun et al 2005), Quinn et al (2010) concluded from a qualitative study investigating client and patient views that regular unsupervised exercise may help to meet therapeutic physical outcomes among people with HD as well as people with PD. As an approach to facilitating this Khalil et al (2012) developed an exercise programme on DVD for home use and investigated the experience of people with HD in carrying out the programme

(unsupervised) through exercise diaries and semi-structured interviews. Caregiver commitment was found to be a key factor in adherence to the programme.

A self-management rehabilitation programme for people with PD included taught mobility and speech exercises, activities of living training on specific tasks, for example turning in bed and dressing, and group discussions on a range of topics such as talking on the phone and preventing falls (Tickle-Degnen et al 2010). Tickle-Degan et al (2010) concluded from their randomised controlled trial (n=115) that whereas self-management programmes tend to be provided for people after significant physical deterioration, greater benefit can result from engagement with the activities in early to middle PD stages.

A novel internet-based programme aimed at facilitating cognitive behaviour therapy-based self-management for fatigue occurring with multiple sclerosis was reported to have demonstrated significant improvement in anxiety and depression contributing to fatigue, in a small preliminary trial (Moss-Morris et al 2012). Qualitative data indicated that most participants found the programme acceptable and helpful in that they came to understand their fatigue better and to find ways to control it.

Mountain and Craig (2012) identified twelve topic areas for self-help programmes based on qualitative interviews with five people with dementia and five carers. Having help to understand and rethink dementia, living with dementia (by establishing routines and learning memory strategies) and keeping mentally well (by recognising and overcoming depression and learning strategies to manage anger and anxiety) were among priorities for participants.

Techniques to help self-manage frustration and fatigue, pain and isolation, exercising, communicating, use of medications, nutrition and how to evaluate new

treatments are taught as part of a ‘chronic disease self-management programme’ (Hirsch et al 2011). Sessions involve participants in developing their own action plans through discussions and problem-solving activities. Relevance of this approach to professionals engaged with people with neurological conditions more broadly has been suggested (Whitehead 2011).

The body of literature concerned with self-management in neurological disease and specifically HD is relatively new and limited but the growing interest in investigating strategies to empower this group is encouraging. Training in activity of living tasks, strategies to limit the impact of symptoms, innovative exercise programmes and use of information technology resources such as internet and DVD to facilitate unsupervised therapeutic activity seem promising avenues for exploration.

A view that researchers and professionals should seek to understand and consider patient and family member perspectives in striving to alleviate the impact of disease experienced as losses is well represented in research focussing on HD (e.g. Ho and Hocaoglu 2011; Aubeeluck and Buchannan 2007). De Boer et al (2007) make the same point with regard to care for people with dementia. Kouzoupis et al (2010) reviewed literature to consider a psychosocial perspective of the impact of multiple sclerosis (MS) and suggest that a narrow, medical view of the condition and of chronic disease generally is often taken by professionals who frequently ignore family members and leave them to feel isolated. They stressed the importance of including family caregiver perceptions of the impact of chronic illness.

2.4 Responses to the experience of motor symptoms associated with HD

HD presents as a complex movement disorder characterised by chorea coexisting with bradykinesia (Thompson et al 1988). More recently Gudesblatt and Tarsy (2011) have suggested involuntary movement as the most common initial symptom of HD and 'restlessness' as the most visible motor sign. As the disease progresses motor impairment becomes increasingly proximal. Dystonia, postural imbalance and tics as well as oculomotor problems are common features but chorea is more noticeable to casual onlookers than more subtle but potentially disabling effects (Gudesblatt and Tarsey, 2011). It is reported that many people with HD are untroubled by chorea personally and that the tendency for health professionals and caregivers to wish the symptom to be treated can be misplaced (e.g. Phillips et al 2008; Thompson 2009 in Stevens et al 2009, p156-163). Quinn et al (2013) indicate that dystonia may occur in up to 90% of people with HD and share the view of Phillips et al (2008) that this can be more disabling than chorea.

However Zeef et al (2011) cite examples of severe and disabling chorea and Poon et al (2010) report that severe chorea can cause social embarrassment, impaired ability to carry out activities of living and suggest that patients experience increasingly extensive disability as chorea becomes more pronounced. Ho and Hocaoglu (2011) found in a qualitative semi-structured interview study examining patient perspectives (n=80) that problems associated with physical function that included chorea were a more dominant concern in mid stages of the disease than were concerns associated with other identified themes such as cognition, emotion, and social aspects.

Bradykinesia is a commonly disabling feature in HD (Phillips et al 2008) and is one of the cardinal motor symptoms of PD along with rigidity which is also seen in

advanced HD, (Quinn et al 2013), resting tremor and postural instability (Haahr et al 2010). As is true for some people with HD patients with PD experience movement disorder as a predominant feature of their condition and consequently have difficulties with tasks such as reaching, grasping and fine motor coordination (Quinn et al 2013). In both conditions the motor disorder occurs in conjunction with psychiatric and cognitive symptoms (Quinn et al 2013). Much of the focus of Health Care Professional (HCP) interventions for HD is on the control of these motor symptoms.

The only medication demonstrated to be consistently effective in the treatment of chorea is tetrabenazine (TBZ) (Huntington's Study Group (HSG) (Tetra-HD) 2006, Frank et al 2008) and in the United States TBZ remains the only pharmacological treatment for chorea in HD approved by the Food and Drug Administration (Videnovic 2013). Tetrabenazine was also associated with improved clinical global impression scoring (HSG Tetra-HD 2006). The Huntington's Study Group suggests that apprehension among physicians regarding TBZ as a drug of choice for the treatment in chorea in HD is due to reported serious adverse events in the treated group, but not in the placebo group (HSG Tetra-HD 2006). The concern may be unjustified: the review for this thesis found no convincing evidence to inform conclusions regarding the causality of these events. However some concern has been expressed regarding dosage where TBZ is prescribed with antidepressants such as Paroxetine and Fluoxetine. TBZ is metabolized by an enzyme in the body known as Cytochrome P4502DS (CYP2D6) and these antidepressant drugs inhibit CYP2D6 (Suchowersky 2013). Armstrong and Miyasaki (2013) agree with Suchowersky's assertion that for patients with HD taking these antidepressants a TBZ dosage should be reduced by 50% but add that whether this advice is adopted clinically is unknown.

Prior to availability of TBZ neuroleptic medications were used to reduce chorea in HD but as these were strongly associated with extrapyramidal effects it is argued that TBZ offers a substantial improvement to quality of life (Hayden et al 2009).

There is evidence of a negative impact of chorea on quality of life whether resulting from HD (Carlozzi and Tulski 2013) or from other causes such as unwanted effects of treatment for PD (Manson et al 2012). This tends to be related to associated mobility impairment (Carlozzi and Tulski 2013) and social embarrassment resulting from chorea in HD and can be justification for treatment with TBZ (Cha 2007). This seems to support the view by Hayden et al (2009) of the potential for TBZ to improve quality of life in HD but counter concerns that TBZ may aggravate psychiatric symptoms including depression (Novak and Tabrizi 2010) and that embarrassment and social impact of chorea is often more a problem for family members and others than for the patient (Carlozzi and Tulski 2013) and this should be considered in any decision-making process about whether treatment with TBZ is appropriate. It should be added that a recent study found TBZ to be effective in reducing chorea with no increase in depression among participants who were taking antidepressants although the authors acknowledge the trial was small and larger trials are needed (Dorsey et al 2011).

Patient satisfaction with the benefits of the drug and cost effectiveness were reported in a small questionnaire survey involving 23 respondents with HD, 39 with Tourettes syndrome and 34 with tardive dyskinesia despite reports of unwanted effects by 82% of participants including drowsiness (54%), slow movement (32%), restlessness (14%) and depression or mood change (12%) (Palao et al 2010). Generally trials of the effectiveness of tetrabenazine for the treatment of Huntington's disease were restricted to investigating movement disorder, particularly chorea and while some

studies such as a recent open label withdrawal pilot project (Fekete et al 2012) measure broader outcomes including cognition, depression, behaviour and other functional scales none found in this review investigate impairment or caregiver outcomes. The limited body of research concerned with wider aspects of the impact of HD and of the effects of the only treatment for the disorder that is demonstrated as effective in rigourous randomised controlled trials emphasises the need for more studies that explore patient and caregiver and other perspectives to enhance understanding of subjective experiences of living with HD and of what is helpful towards doing so.

Other drugs evaluated for their efficacy in treating HD were Amantadine, Apomorphine, Aripiprazole, Atomoxetine, Baclofen, Cannabidiol, Clozapine, Coenzyme Q10, Creatine, D-alpha-tocopherol, Donepezil, Ethyl-EPA, Fluoxetine, Idebenone, Ketamine, L-acetyl carnitine, Lamotrigine, Latrepirdine, Minocycline, Mixed oral unsaturated fatty acids, Modafinil, OPC-14117, Piracetam.Prodipodine, Remacemide, Riluzole, Rivastigmine, Sulpiride, Terguride and Tiapride. Although some of these drugs have shown initial promise none of the studies evaluating their efficacy have had sufficient numbers of participants, duration of follow-up, or methodological quality to ensure that the results are robust and reliable enough to be able to claim efficacy (Mestre and Ferreira 2012).

The findings of some fairly recent studies offer hope for the future. The ‘HART’ study (n=22) (Tedroff 2010) and the ‘MermaiHD’ study (n=437) (Garcia de Yebenes et al 2011) have generated optimism regarding Pridopidine for improving motor function in HD. A trend towards improvement in affective symptoms was reported in a small scale study of Pridopidine (Lundin et al 2010) but none of these

trials have so far demonstrated significant improvement on any primary outcomes. The potential for Pridopidine to improve life for people with HD has yet to be established.

Potential value of some medications for some HD patients cannot be ruled out. Apomorphine may benefit carefully selected individuals with HD (Vitale et al 2007; Albanese et al 1995) but the evidence for this is low.

The conflicting findings from the various trials of effectiveness of ethyl-EPA are tantalising with one trial involving 316 people with HD showing no significant effect on motor function (Trend-HD 2008) while another smaller study involving 30 patients with advanced HD demonstrated significant reduction in cerebral atrophy rate particularly in localised brain regions associated with movement disorder in HD (Puri et al 2008). Ethyl-EPA looks set to remain a focus for investigation.

Zeef et al (2011) point out that so far patients included in studies of deep brain stimulation (DBS) for HD have been selected based on having motor problems with minimal emotional disturbance. This is in keeping with practice in studies of DBS for Parkinson's disease where dementia and depression are excluded due to poor outcomes for patients with these co-morbidities (Fasano et al 2010). For this reason while encouraging, results cannot be generalised. Establishing DBS as a treatment option seems set to be a long-term future project, with some promise for a limited group within the HD population.

The only study to convincingly demonstrate efficacy for neural transplantation for HD is that carried out in Créteil, France (Bachoud-Lévi et al 2000[a & b], Bachoud-Lévi et al 2006). The study aims and efficacy demonstrated were concerned not only with movement disorder but also cognitive deterioration associated with HD. With the

low number in the trial and only three of five patients showing improvement, and the potential for bias due to the non-blinded design, as Dunnett and Rosser (2007) advised, cautious interpretation is appropriate even for this most rigorous of trials so far undertaken. As they suggest while some optimism for the future may be appropriate, reliability and generalisability have not been established.

In any case it should be noted that foetal neuron transplantation, while useful in research seeking to establish proof of concept, can never become widely adopted as a treatment. The ethical 'minefield' of using foetal tissue aside, practical difficulties in supply would be insurmountable as Dunnet and Rosser point out (2007). Whether the future will bring to prominence embryonic stem cells, foetal neuronal precursors or adult stem cells is a debate beyond the scope of this review.

Bilney et al (2003) and Quinn and Rao (2002) point out that the low-level evidence that exists does support the use of physiotherapy for treating balance and muscle strength problems and ability to carry out daily activities. Busse and Rosser (2007) highlight the need for valid and reliable assessment tools to aid evaluation of the effectiveness of rehabilitation interventions for HD. In PD a recent Cochrane systematic review (Tomlinson et al 2012) concluded that physiotherapy provides short term benefits in this disease, and that little difference was shown in treatment effects between a wide range of techniques. A comparable body of evidence is needed to evaluate similar therapeutic input for people with HD.

One within-subjects design study (Zinzi et al, 2006) (n=40) examined the effect of a three-week intensive therapy programme for people with HD. The result of 'no change' between baseline and the follow-up two years later was proposed to demonstrate maintenance of function and motor performance. This pilot study needs

to be followed with a study that has a comparator group, larger numbers, uses independent blinded assessment and reports on whether or not people who miss some sessions benefit from their partial involvement in the programme.

Conflicting results suggest that the use of ECT to treat chorea in HD cannot be supported (Leroi and Michalon 1998; Beale et al 1997; Lewis et al 1996). ECT is also discussed below in the context of treatment for psychiatric HD symptoms.

While there are many pharmacological and non-pharmacological interventions that may have an important place in reducing motor symptoms in HD, evidence in support of this is lacking and the only treatment for which there is robust evidence- Tetrabenazine for chorea- does not have support for a positive effect on the wider impact of the symptoms on patients and their caregivers. A lack of evidence does not mean that interventions are not effective and therefore robust research in a range of therapies aimed at treating motor symptoms in HD utilising broad outcome measures must be supported.

2.5 Responses to the experience of psychiatric symptoms associated with HD

Psychiatric disorders, particularly major depressive disorder have been demonstrated to be a common experience among people with HD (Redeeker et al 2012). Uncertainty surrounds the incidence and prevalence of psychosis in HD (Crooks and Rickards 2012). In their data-mining study involving data sets from 998 participants, prevalence was 6.5% and incidence was 2.8 new cases per 100 patients per year (Crooks and Rickards 2012). However these authors concluded that further validation of the measure used (UHDRS) to identify hallucinations and delusions is necessary before these results can be considered meaningful.

Fifty two consecutive people with HD recruited from a clinical research programme at two US locations were interviewed using standardised measures of psychological impairment (Paulsen et al 2001). In 98% neuropsychiatric symptoms were identified with dysphoria, agitation, irritability, apathy and anxiety most prominent.

Estimates of the prevalence of depression in people with HD vary from 9% to 63% and several sources suggest an occurrence of nearly 50% (Paulsen et al 2005[b]). De Souza et al (2010) highlight difficulties in diagnosing depression in HD because indicators such as weight loss, sleep problems, loss of concentration and fatigue are inherent in HD. This potentially leads to incorrect diagnosis of depression in people who are showing the first signs of HD. Additionally the diagnosis of depression can be missed as a co-morbidity of HD where the symptoms of depression are readily dismissed as core features of HD. Ho et al (2004) identify depression in HD as associated with impaired quality of life and Paulsen et al (2005)[b] cite evidence to suggest depression and other psychiatric symptoms can be the presenting complaint and may be present up to 20 years prior to a diagnosis of HD.

The experience of people with HD regarding psychiatric symptoms is arguably similar to that of people with other degenerative neurological conditions. In a study comparing the frequency of neuropsychiatric symptoms in people with PD with and without dementia to patients with dementia with Lewy bodies (DLB), delusions and hallucinations occurred in all three groups, most commonly in DLB, and least in PD without dementia (Aarsland et al 2001). In all three groups the presentation of these symptoms was similar. The negative impact of psychiatric symptoms on the quality of life of people with PD is well documented (e.g. Barone et al 2009, Gómez-Esteban et al 2011). Anxiety disorders are reportedly common but under-investigated

in PD and in a case-series study (n=79) a 14% occurrence of comorbid anxiety and depression was observed (Dissanayaka et al 2010). Depression is said to affect up to 50% of people with PD (similar to estimates of depression in HD) and to be associated with poor outcomes for individuals and their families (Menza et al 2009). One study found a higher prevalence of PD among hospital patients who had depression than among those without depression (Miu and Chan 2011). They demonstrated that depression was associated with a higher prevalence of functional decline and mortality. Depression in PD is regarded as one of the most disabling aspects of PD (Hemmerle et al 2012).

The prevalence of anxiety and depression in association with HD is greater than among the general population (Dahl et al 2009). This has also been found in other neurological conditions such as MS and PD. Burns et al (2013) identified an association between exacerbations of MS (both pseudo and confirmed) and anxiety and depression. Giordano et al (2011) noted an association between anxiety and depression and found anxiety to be prevalent and persistent during the period leading up to and around the time of diagnosis in MS patients. Levels of anxiety were not influenced by the diagnostic result and Giordano et al (2011) concluded that recognition and intervention for anxiety in the early period may reduce the likelihood of long term depression. Depression is established as a common complication of PD (Reijnders et al 2008) and anxiety in PD is reported to be under-recognised and undertreated (Pontone et al 2009).

In a systematic review Mitchell et al (2010) found that across a range of neurological conditions including HD, PD, MS, stroke, epilepsy, head injury and motor neuron disease, cognitive impairment often occurs and is frequently overlooked by

clinicians and suggest that earlier recognition might reduce the negative impact of disease on quality of life. Mitchell et al (2010) concluded that in spite of awareness of the importance of profound cognitive decline amounting to dementia the broad spectrum of impairment including subclinical, subtle cognitive changes are increasingly becoming recognised as significant factors affecting quality of life in all of these diagnoses.

An association between behaviour problems in HD and earlier admission to residential care has been demonstrated (Fisher et al 2012). In a considerable minority of people with PD challenging behaviour poses problems for caregivers (Leeman et al 2012). A systematic review and meta-analysis investigating the prevalence of behavioural problems in multiple sclerosis concluded that behaviours such as aggression, apathy, euphoria and lack of insight are problematic and under-recognised in this condition (Rosti-Otajarvi and Hämäläinen 2013). Behaviours that lead to distress among family members frequently occur in dementia suggesting that caregivers would benefit if these symptoms were targeted and addressed (Gitlin et al 2010).

It is clear then that psychiatric symptoms and related problems with management of behaviour cause distress and disability across diagnoses but while hallucinations and delusions have been found to be the strongest predictors of admission to nursing homes in PD (Aarsland et al 2000; Goetz and Stebbins 1993), physical factors have been claimed as more commonly the reason for institutionalization in HD (Rosenblatt et al 2011). Fisher et al (2012) dispute this however, and refer to potential bias acknowledged by Rosenblatt et al relating to high prevalence of psychosocial and behavioural problems among participants lost to follow-up as a

possible explanation for underestimating the relationship between psychiatric problems in HD and institutionalisation. Reduction or delay in institution admissions associated with community interventions to improve psychosocial and behavioural factors such as counselling for carers, behaviour management strategies and case management has been demonstrated with regard to dementia (Brodaty et al 2003, Mittelman et al 2006) and it is suggested that similar approaches could alleviate caregiver duress and avoid unnecessary admissions to long term care in HD (Fisher et al 2012).

Currently there appears to be no evidence to support physicians looking for effective pharmacological management strategies for psychosis in HD (Mestre and Ferreira 2012). One case study (n=1) evaluated cognitive behaviour therapy (CBT) for effectiveness in treating depression and anxiety relating to a positive genetic test for HD (Silver 2003). The subjectivity within the discussion of reported benefit for this individual does not indicate that CBT is not effective for HD, but currently there appears to be no substantial evidence in support.

The small number of case studies examining the effect of electroconvulsive therapy (ECT) for HD (Leroi and Michalon 1998; Beale et al 1997; Lewis et al 1996) are accounts of despair demonstrated by some harrowing suicide attempts. Although the impact of HD is frequently described as devastating, these extreme cases are fortunately atypical. ECT has been seen as an option worth trying despite uncertainty regarding effectiveness. The resulting evidence is little more than anecdotal but appears to be the best available. The generally favourable reports regarding treatment for depression in HD are likely to be valuable support for discussion in cases where every other attempt to relieve despair have failed.

There is no good quality evidence to help inform clinical teams of the best pharmacological option for the treatment of anxiety and depression in HD. Latrepirdine shows some promise with regard to improved cognition on the mini mental state examination, although no benefit compared to placebo was demonstrated on the UHDRS or the Alzheimer's Disease Assessment Scale-cognitive subscale (ADAS-cog) (Kieburtz et al 2010).

One randomised controlled study examined the effects of multisensory stimulation environment (MSE) on rehabilitation, behaviour and mood disturbance and motor assessment (Leng et al 2003). The small study (n=10) found an unsustained benefit of improved mood and behaviour in the treatment group.

Three studies sought to explore or evaluate music therapy as a treatment for HD (Magee 1995; Davis and Magee 2001; Daveson 2007). Improvisation techniques were reported to benefit people in early to middle stages, and more structured and familiar music benefits people in advanced stages of HD (Magee 1995). Music therapy was said to bring about improved mood associated with positive changes in levels of interaction and to provide an alternative means of communication for people with HD (Davis and Magee 2001).

2.6 Responses to the experience of stigma associated with HD

Historically social stigma associated with HD has led many families to keep the diagnosis a secret and stigma continues to add to suffering (Wexler 2010). According to Wexler although enlightenment through scientific knowledge can reduce stigma it can also exacerbate the problem as occurred when science underpinned the eugenics movement; the view that the problem of HD might be eradicated through ensuring people with HD do not procreate, may not yet be

extinct. Nevertheless stigma is generally understood to result from ignorance (lack of knowledge of the condition) (Wexler 2010). Hence the value of knowledge families have gained through their experiences, a principle adopted in the 1977 by the USA Congressional commission for the Control of Huntington's Disease which had extensive input from family members and along with newly forming lay groups was reportedly instrumental in influencing research to avoid seeking eugenic solutions (Wexler 2010).

Apart from the issue of social stigma, HD is a complex condition that requires knowledge in a variety of areas in order to optimally manage it. Patients and their carers gain knowledge of their condition through a variety of sources, and familial expertise in this genetic disorder is not to be overlooked (Simpson and Rae 2012). Simpson and Rae suggest that this point has been embraced by the European Huntington's Disease Network (EHDN) Standards of Care working group whose aim is to develop internationally approved guidance for HD care based on understandings of the condition informed by family expertise (Simpson and Rae 2012).

A lack of knowledge of HD among health professionals has been identified as an issue of concern in literature relating to HD as this can negatively impact on the organisation and delivery of care and services. Unintentional stigmatisation may result from a lack of knowledge or 'ignorance' (Wexler 2010). Dawson et al (2004) highlight a need for disease-specific training among staff in hospitals and residential care settings providing for people with HD and found that poor knowledge of the condition among staff caused a lack of trust in the standard of care. This point has been raised with regard to MS by Schneider and Young (2010) who found a need for physicians to have a greater knowledge of MS, so that they were able to give clear

guidance. The commonly reported inability of physicians to do so was a cause of frustration among participants in that study.

One paper investigated online support for people affected by HD (Coulson et al, 2007). The authors undertook a content analysis study of messages on one social network site concerned with HD. The authors highlighted the potential value for health professionals of using the facilities to understand discrepancies between evidence-based practice and consumer beliefs, behaviour and expectations. Of course this does pre-suppose expertise on the part of the HCPs informing the debates. Concern among professionals for the wellbeing of people who have experienced a stroke or transient ischaemic attack (TIA) and do not seek medical attention led to a study to determine whether people who seek information about these conditions online can be targeted (Kim et al 2012). An advertisement placed on a website designed to be reached by people searching for information on TIA and stroke led to 251 (1%) of over 25,000 visitors over 122 days completing an online questionnaire and phone contact between professionals and people identified as consenting to and in need of professional input. Although Kim et al (2012) were concerned with providing emergency medical intervention the study does demonstrate a tendency for some people with neurological symptoms to seek information via the internet rather than direct initial contact with health professionals and there may be implications for the importance of providing good information online for people seeking to gain knowledge about degenerative neurological disorders.

2.7 Conclusions regarding responses to experiences of living with HD

There are then many aspects to the experience of living with HD and despite some unique characteristics of the condition it appears that to some extent people with other neurological disorders also face similar challenges. Research activity concerned with effectiveness of interventions for HD has been most intensive with regard to the problem of the movement disorder and has resulted in identifying the only treatment for any symptom so far demonstrated as effective, Tetrabenazine. Interventions that are frequently adopted as responses to other aspects of the experience of HD may be effective also but to date substantial evidence for this is lacking. The seemingly disproportionate engagement in research concerned with movement disorder (Gudesblatt and Tarsey 2011) compared to investigations of effectiveness of responses to non-motor problems emphasises the importance of being guided by the perspectives of individuals with HD and their families. It can be argued that they are in a far better position to identify the aspects of HD that have the greatest impact on their lives and so their opinions should drive the prioritisation of research into interventions.

This need to understand subjective experience of those who live with long term neurological conditions including HD is increasingly appreciated and some of the methods adopted to enhance insight in this area are considered below.

2.8 Methods used to understand living with neurological disorders

The studies considered in this review that have reported on the prevalence of disease or of symptoms associated with specific diagnoses adopt a quantitative survey design as is appropriate for these purposes (Abramson and Abramson 2011) and those that examined the effectiveness of interventions generally used a quantitative experimental design which is also suitable to meet the aims of such studies

(Abramson and Abramson 2011). The studies investigating the experience of lving with the conditions, the impact on quality of life and any that seek to aid understanding of subjective perspectives that are discussed in the review used qualitative approaches and these methods are much more suited to questions of this nature (Bowling 2009). The body of knowledge is informative with regard to understanding of what it is like to live with HD, the range of experiences and problems that are associated with the condition and of what is known so far about the effect and experience of the many interventions aimed at alleviating some of the specific challenges that HD poses. None of the work within the reviewed literature investigates perspectives of HD patients, their caregivers and the health professionals providing their care and treatment about what is helpful towards living with HD in a way that would enable participants to make a broad range of suggestions including for example, therapeutic interventions, social or politico-economic factors or any other aspect they might consider to be important. This is an identified gap that this thesis is intended to address. The topic area concerns subjective opinions drawn from experience of HD and therefore methods that have been used in qualitative studies discussed in the review should be considered in the process of determining the approach most suited to answering research question.

One common approach to gaining insight into patient and/or caregiver perspectives about some aspect of living with a neurological condition is through questionnaires. The development of suitable questionnaires aimed at understanding patient and caregiver quality of life with regard to Huntington's disease is ongoing (Hocaoglu et al 2012; Aubeeluck et al, 2012). Recent examples of this approach in other neurological conditions include a study of the experience of fatigue in PD (Beiske et al, 2010); a self-report survey examining employment changes and the importance of

symptom management in relation to living with multiple sclerosis (Simmons et al 2010) and identifying personal factors that influence the experience of living with motor neurone disease (Ng and Kahn 2011).

However, a questionnaire approach is suited to studies in which the questions are 'straight-forward and simple' and less suited to addressing complex issues (Bowling 2009) such as the broad question that this current study poses: 'What helps people with HD live with their condition?' On the other hand, self-reported questionnaire methods, especially postal, have the benefit over interview approaches of being more likely to reduce researcher bias through social engagement that may lead respondents to give answers they perceive as pleasing to the researcher (Bowling 2009).

Qualitative interview methods have been adopted for example to understand the subjective family experience of stigma associated with Alzheimer's disease (Werner et al, 2010) and to explore the experience of living with a partner with Parkinson's disease who is undergoing deep brain stimulation treatment (Haahr et al, 2013). Whether the interview format is structured or unstructured, this approach does have the advantage of enabling the interviewer to explore any uncertainties and to prompt if necessary, reducing the likelihood of missing data particularly if the interviewer is friendly and motivating. Bowling (2009) suggests that qualitative interviews pose a low burden on respondents as they do not need writing skills and can speak naturally.

Quinn et al (2010) interviewed patients with PD and HD and suggested this approach had an advantage over focus groups in allowing participants the convenience of being more freely able to choose the time and location to take part. However they did also use a focus group method in their study examining views on exercise

programmes for these conditions and indicated that focus groups are compatible with other qualitative data collection methods and often result in a broader scope of topics arising as a result of engagement in discussion. For some participants focus group activities may provide a greater sense of security than interview methods (Webb and Doman 2011).

Mixed-methods (qualitative and quantitative) are becoming increasingly recognised as valuable in health research because of the capacity to draw on the strengths of each (Curry et al 2009). Curry et al (2009) point out the potential to identify rich subjective data via a qualitative component, and examine this further through quantitative means. This has appeal with regard to the current study because it could accommodate exploring what helps people live with HD using qualitative methods akin to focus group and interview approaches, and then consideration of what is most important through a quantitative component.

2.9 Selection of the Concept Mapping method

To answer the question: 'What helps people with HD live with their condition?' the traditionally dominant positivist approach to the acquisition of knowledge was rejected due to both the nature of the question and also to the primary investigator's personal and professional history of involvement with the topic area. Positivism assumes an external and concrete reality and requires that research is concerned with objective methods designed to uncover truth (Brown 2003). This study is concerned with subjective views and opinions about what participants believe is helpful and with how they conceptualise and prioritise the factors perceived to help. There is no concrete 'truth' in the positivist sense to uncover.

Interpretivism views the social world as actively constructed by human beings, and the qualitative methods generally associated with it are broadly concerned with investigating how people make sense of their contextual situations and circumstances (Parahoo 1997). This seemed a closer fit to the current inquiry, and a number of qualitative approaches associated with the interpretivist paradigm were considered.

For example, a grounded theory approach could involve gathering statements or recording dialogue from individuals and/or groups, and constructing a theory from a thematic analysis. The current study does seek to be inductive rather than deductive, that is, to draw conclusions derived from data rather than test any hypothesis. But the aim is to represent opinions of participants, not to investigate the social processes that may underlie how the opinions came to be, or to build a theory of how these processes work.

Another approach that initially seemed as though it might be suitable for this study was a 'Delphi study' which it was felt could have been adopted to obtain and analyse rich, contextual qualitative data (Mason 1996). It is a method that involves engaging in discussions with participants in natural settings (Cresswell 1994) and would facilitate consideration of what helps people to live with HD by distilling the judgements of carefully chosen experts (Adler and Ziglio 1996), in this case the people who do live with the condition, the caregivers and the health professionals. However, the tendency to force a 'middle of the road consensus' as a way of dealing with views that might be considered extreme by most participants, is inherent in Delphi studies (Barnes 1987) and would be a disadvantage here as there is a need to represent the whole range of views in this current project.

A phenomenological approach was also considered because it would provide insight into the lived experience of participants. Pringle et al (2011) for example discuss the challenges of using phenomenological approaches to exploring the experiences of people who have survived a stroke. Qualitative methods involving focus groups or individual interviews are examples of means for achieving this kind of insight. But it was felt that the aims of this current study could not be entirely met by exploring the lived experience of being or caring for someone with HD. The purpose was to obtain opinions about what helps, and to learn how people conceptualise and prioritise factors that are perceived to be helpful.

Concept mapping is designed to obtain statements of opinion, prioritise them and graphically represent the way that participants conceptualise the relationships between the statements. The concept mapping method was selected for reasons explained below.

The concept mapping method involves participants in a brainstorming activity in which statements are generated that capture answers to the research question. This tends to be done in groups arriving at the statements through discussion of aspects of the problem being considered but the method allows for some participants to generate statements individually (Kane and Trochim 2007). A statement reduction process by the researchers eliminates duplicate statements and those that do not answer the research question. The reduction process also in some cases involves creating one single statement that the steering group and validation group (these groups are defined in the next chapter) agree sums up several original statements. The developers of the concept mapping method (Trochim and Linton 1986) suggest that it tends to be possible to capture the points arising from brainstorming sessions within no more that 100 statements. In a separate activity, participants organise the

statements (each printed separately onto a numbered card) into ‘clusters’ or groupings indicating their individual subjective view of which statements seem to belong together for some reason. Then the participants each prioritise the statements by placing each into one of five stacks numbered one to five, five being the stack containing the perceived most important statements and stack one containing the perceived least important statements. The stacks should be distributed so that each contains a similar number of statements. The data collection process is explained in more detail in Chapter Three.

Specialised computer software (‘Ariadne’®) is used in the analysis to determine how often participants placed any two statements together in a cluster, and to calculate a mean priority score for each statement based on which priority stack (1-5) each participant placed each statement. The mean priority score for each cluster could then be calculated.

The reason for analysing the way the statements are grouped by participants into clusters is to be able to derive more meaning from the data than would be achievable through a list of stand-alone statements. Kane and Trochim (2007) point out that creating graphic maps is an inate human activity that allows us to make sense of where we are and what choices we may have about moving from one place or situation to another. The clustering of statements onto a map facilitates a view of how the thoughts of groups of participants are organised, and how in this case some statements are perceived to relate closely to each other while others may be more distant, separate issues. The reason for prioritising the statements that will be grouped into clusters is to enable calculation of mean priority scores to determine which clusters are perceived by the participants as a group to be most important. It is

also valuable to note which particular statements stand out as having high importance among participants.

Based on a study involving four GP groups from different demographic areas, concept mapping was reported to be a very useful method with the ability to capitalise on the advantages of both qualitative and quantitative research approaches (Southern et al 1999). Ryan et al (2001) found it similar to using focus groups but attribute concept mapping with the quality of ensuring that all participants have an equal opportunity to express opinions, avoiding what they describe as the problems group dynamics can cause. Trochim and Linton (1986) suggest that the method is suited to informing decision-making and prioritising, which fits well with the aims of this current study. The difference between informing decision-making and helping to reach decisions (the latter lending itself towards a consensus method) should be noted. These factors form the rationale for the choice of this method. Qualitative data is gathered in the form of statements agreed by participants that answer the question (in this case 'what helps people with HD live with their condition?'). Quantitative analysis is used to prioritise statements enabling the consensus among participants about which statements are more important than others, and also to calculate the frequency with which participants group particular statements together so that the software can present a visual map of 'clusters' of statements, demonstrating how statements are conceptualised into groupings. Because both qualitative and quantitative elements are inherent in the process Trochim and Linton (1986) described their approach as 'mixed methods'.

Applications of concept mapping

The term 'concept mapping' here refers to the structured method developed by Trochim and Linton (1986) as opposed to methods of setting out ideas designed to

be used by individuals, as the term is often used (Trochim and Kane 2005). Since its introduction the approach has been used in a diverse variety of social research projects beyond a direct health context.

For example Ridings et al (2008) used concept mapping to steer a community building project by a large non-profiting organisation with a grant provided to benefit African Americans in a metropolitan area of Chicago. Phang and Lee (2009) studied the experience of social support among working mothers. Community relations in Northern Ireland were studied by Knox (1995) using this method. Its use towards steering projects, investigating experience and evaluating policy demonstrated in these three examples highlights the adaptability of concept mapping.

Health research adopting concept mapping embraces a broad range of topic areas such as beliefs about chronic back pain held by patients (Knish and Calder 1999), determining an optimum treatment location (home versus centre) for community rehabilitation (Barker and Ziino 2010) understanding patient perspectives of breast cancer care (De Kok et al 2007) and facilitating job retention for chronically ill employees (Haafkens et al 2011).

Trochim et al (2004) used the method to inform a state-wide health improvement initiative for one USA state with a need to quickly and appropriately disseminate unanticipated funds arising from a tobacco settlement agreement. The method was considered particularly suitable due to the ability to facilitate the complexity of the undertaking, the speed with which results were needed to help reach decisions and the importance of incorporating multiple stakeholder perspectives (Trochim and Kane 2005).

De Ridder et al (1997) used concept mapping to elicit perspectives of patients on ways of coping with illness and with the healthcare system. One of the co-authors (Peter Severens) went on to develop the 'Ariadne®' software used for analysis in this thesis. The experience of depression among college students was studied by Daughtry and Kunkel (1993). Daughtry and Kunkel (1993) found concept mapping particularly useful in helping to clarify the constituent elements and underlying structure of depression as experienced by the college students.

In a study by Valentine (1989) views of three participant subgroups (nurses, patients and nurse theorists/ researchers) were presented in separate concept maps to depict how 'caring' was perceived in a nursing context. Another study comparing subgroup perceptions was carried out to shed light on differences between patient and medical specialist perspectives of preferences of aspects of care in relation to quality of care with regard to chronic diseases, namely chronic obstructive pulmonary disease, rheumatoid arthritis and diabetes mellitus (van Der Waal et al 1996).

Summary of advantages of utilising concept mapping for this question

The concept mapping method then can accommodate study into wide-ranging health and social care issues where stakeholder views are sought to establish what is important and how the identified factors can be conceptualised and prioritised to inform decision-making regarding practice, service provision and policy. The method facilitates handling of complex issues and relatively speedily provides results that can inform decision-making (Trochim et al 2004). Kikkert et al (2006) suggested that because the 'clustering' task enables participants as opposed to researchers to decide which grouped factors emerge from the data and how these 'clusters' of statements are related, interpretation of the qualitative data may be less susceptible than other qualitative approaches to researchers' preconceptions.

Concept mapping is useful when stakeholder perspectives are important to understand (De Ridder et al 1997). The approach can clarify the structure and constituent elements of perceived phenomena (Daughrty and Kunkel 1993) and so for example if 'social inclusion' were identified as helpful towards living with HD it may be possible to see specific components of the concept of 'social inclusion' and how these are organised.

Comparison of perspectives between participant subgroups is supported by concept mapping (van der Waal et al 1996; Valentine 1989) and therefore the intention to compare views of people with HD, caregivers and professionals in the current study can be accommodated. Valentine (1989) highlighted the high level engagement of participants in stages of the process and their tendency to enjoy participation as advantages for the method. A further advantage pointed out by Valentine (1989) that is particularly attractive for the current study is the visual representation of results. It was felt that this could enhance dissemination, enabling people with HD and caregivers as well as professionals to readily understand results when presented at conferences and small group meetings.

Chapter Three
Methods

3.1 Introduction

A mixed methods participatory approach known as Concept Mapping was adopted to enable exploration of the views of participants representing three stakeholder groups: people with Huntington's disease (PwHD), family caregivers (Carers) and health professionals who work with patients (or clients) with HD (HP). A rationale for this choice of method is provided in the previous chapter.

3.2 Preparation

3.2.1 Project Monitoring

Project monitoring was carried out through monthly meetings between the primary investigator and PhD supervisors. Day to day progress was also reviewed through this process.

3.2.2 Steering group

Quarterly 'steering group'meetings involved the above project monitors and also two PhD students, (HF and AM) who were both also using concept mapping methodology in their projects. These meetings informed the study design.

3.2.3 Validation group

Inclusion of people with HD and caregivers in the steering group was intended but discussions with potential candidates led to the conclusion that this was not practical. Instead the principal investigator met with members of a branch of the Huntington's Disease Association at key stages: to agree the protocol; to discuss the ethics

application prior to submission; to validate the process of statement reduction following generation of statements at brainstorming sessions; to discuss and validate (or contest) results; to validate (or contest) draft discussion and conclusions prior to thesis submission. The intention to ensure this 'validation group' included at least two of each of health professionals, people with HD and family caregivers at each meeting was achieved. This 'service user' involvement in research design is in keeping with National Research Ethics Service recommendations (NRES 2011). In addition the draft protocol was summarised in presentations by the primary investigator to health professionals with an interest in HD and/or other neurological disorders with a view to noting comments given as feedback, and where appropriate adjusting the protocol.

3.2.4 Project focus

In line with the concept mapping process the steering group decided on the research question which would form the basis for construction of the concept map (Novak and Canas 2008). This question *'What helps people with HD live with their conditions is…'* was displayed prominently in the room during statement generation and sorting activities to aid participants in remaining focussed on their task.

3.2.5 Ethical Issues

Ethical permission for this project was granted by the NHS National Research Committee, Cambridge 2, on September 25th 2009. The key issue of concern for the committee was safeguarding the rights and wellbeing of people with HD, especially those with advancing disease, who may not have capacity to give informed consent in accordance with the Mental Capacity Act (2005). On the other hand, the committee members were appreciative of concerns raised by the 'validation group'

referred to above, that the views of people with advanced HD are important and that they should not be excluded.

The Research Ethics Committee commended the plan for dealing with this issue. The plan involved identifying a 'consultee' to assist me as primary investigator, in deciding whether a participant without capacity to give informed consent, who indicated willingness to take part, should be enrolled, and whether a person who appeared to lose capacity and who wished to continue taking part should be allowed to do so.

As agreed with the Research Ethics Committee, care was taken to avoid disclosure of factors that might have identified participants in the following way. The reply slips and consent forms and signed information sheets were (are) kept securely in a locked filing cabinet within a locked room within the School of Nursing Sciences at the University of East Anglia. Personal details were held on a university computer with password security, in a document with further password security. Apart from contact details, the personal information recorded for the purpose of analysing data was:

All participants: Gender; date of birth;

People with HD: Length of time since clinical HD diagnosis.

Caregivers: Relationship to the person they care for; length of time caring for a person or persons with HD.

Health Professionals: Name of profession. Number of patients with HD on current client list; length of time involved with care for people with HD.

Personal identities were not made known to others- summaries of results used in any publications ensured anonymity. The forms that contain the data referring to the way

each participant prioritised and clustered the generated statements, contained a code number relating to the personal information held on the consent forms and reply slips.

3.3 Recruitment

The originally planned method for recruitment, agreed by the Research Ethics Committee entailed recruiting three groups that had been identified as key stakeholders with a high level of expertise in relation to the topic (people with HD, caregivers and health professionals) each of between 10-20 participants, accessed through a neurology department database in Cambridge, UK. Trochim (1989) suggests that there is no strict limit concerning numbers of participants in groups. He highlights successful concept generation involving one lone participant, and also groups of 75-80 people. Typically he reported using groups of 10-20 and found this a workable number. Whatever the group sizes, conceptualising is best according to Trochim (1989) when it involves a wide variety of relevant people.

The inclusion criteria for members of the three stakeholder groups, as agreed by the steering and validation groups were:

PwHD: Clinically diagnosed HD confirmed by genetic testing.

Family Caregivers: Identified by the PwHD as his or her primary informal caregiver (unpaid).

Health professional: current experience of providing care, services or treatment for at least one person with HD.

Initially recruiting participants from groups via the Huntington's Disease Association (HDA) seemed a practical option because groups already meet, negating

the need to bring strangers together, find a venue and arrange refreshment facilities. But recruitment from a clinic database was decided on as an attempt to reduce potential selection-bias because people who take part in support groups may tend to self-select and may be unrepresentative of others affected by HD who do not.

Health professionals were recruited from attendance lists from professional meetings with an HD focus, accessed via the European Huntington's Disease Association Network at various locations in England.

The recruiting method needed to be revised after attempts to recruit from the clinic database were largely unsuccessful. An amendment agreed by the Research Ethics Committee allowed in addition to the initial procedure, enrolment of people with HD and their caregivers via branches of the HDA, and of people with advanced HD, caregivers and health professionals at a named specialist Huntington's Disease care facility within a large neurodisability hospital in London. This second part of the amendment was requested as it had been realised that the initial plan seemed unlikely to be very successful in gaining access to participants for whom the context of living with or caring for a person with HD is based in a long-term care setting rather than in the community. Local Research and Development (R&D) approval was obtained to recruit within their specialist HD wards.

The potential problem referred to earlier - that people who attend Huntington's Disease Association branch meetings may not be representative of other people with HD - is acknowledged. It is also recognised that the large specialised long-term care facility is not typical of long-term care facilities caring for people with HD, many of which may be smaller nursing or residential homes, and may not be specialised,

caring for example, for elderly people. Copies of ethics approval documents including the amendment are included in Appendix 3.1.

Potential participants with HD were contacted initially by telephone (prior to recruitment amendment) and those who accepted the invitation were sent a postal package containing a letter of invitation, an information document, a consent form, a reply slip and a second pack labelled 'Carer Pack'. Potential participants were advised not to complete any of the documentation prior to attending the first activity, the brainstorming session. The letter of invitation asked if the person with HD would kindly hand the 'carer pack' to the person they regard as their main carer. The carer pack also contained a letter of invitation, an information document, a consent form and a reply slip. The pack sent to potential health professional participants contained similar documentation (Appendix 3.2).

A follow-up phone call aimed to establish the time and place of the brainstorming session. Signed consent was obtained at the brainstorming session venue, prior to commencing the activity. Participants were reminded that they were not obliged to take part and that they were free to discontinue at any time, without the need to give any reason. In the case of participants recruited via Huntington's Disease Association branches, the material described above was given to the HDA branch leader, in envelopes, to hand to people with HD, who in the same way, were asked to decide who to hand the enclosed 'carer pack' to.

Fig 3.1 Flow Chart- Recruitment method 1:

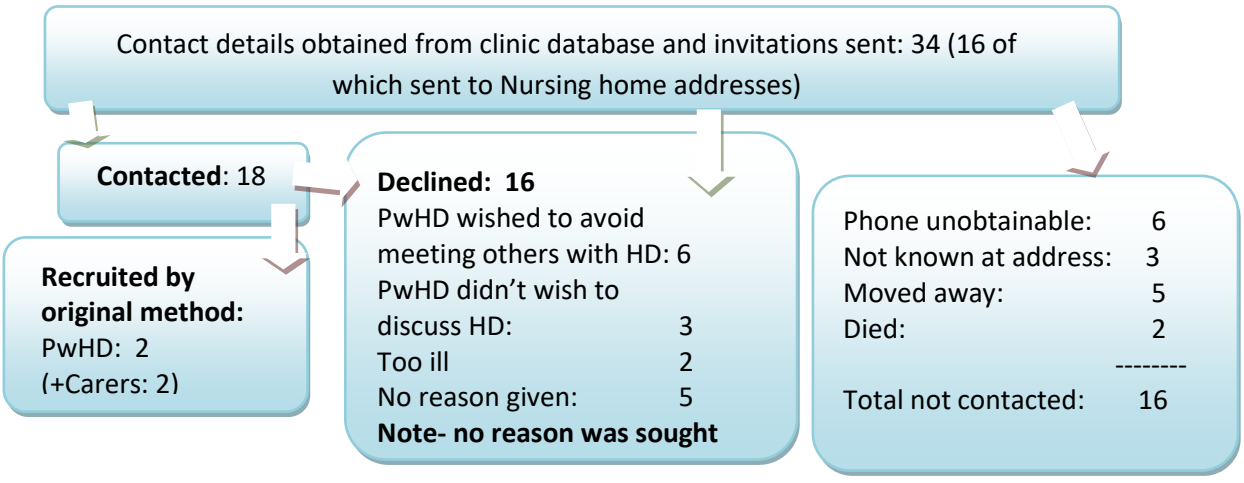


Fig 3.2 Flow chart- recruitment method 2.

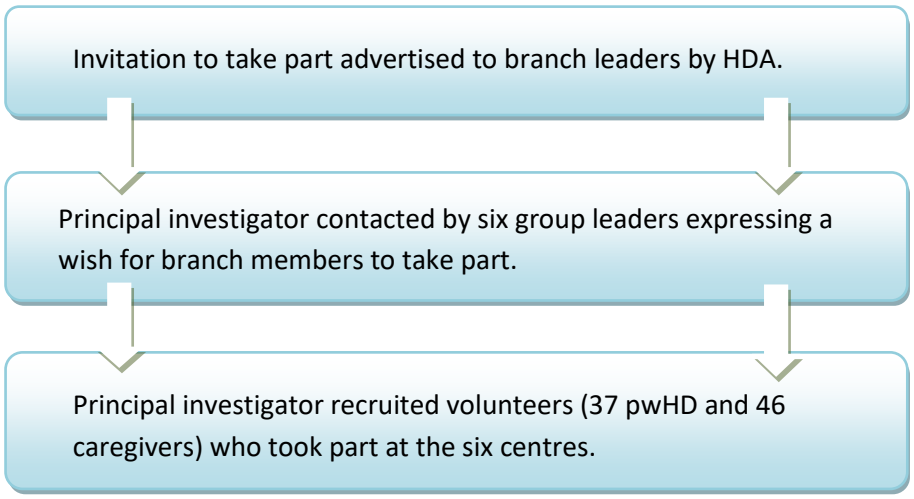
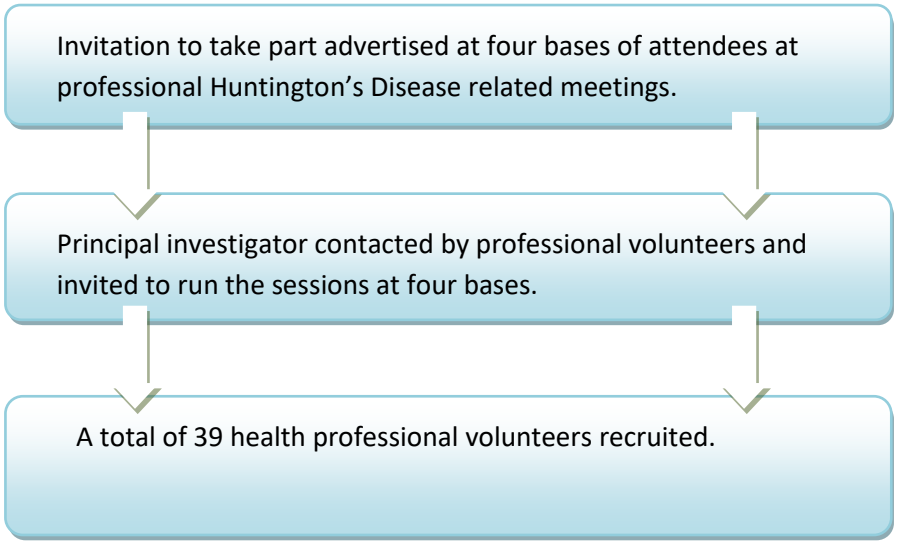


Fig 3.3 Flow chart- Health Professional Recruitment.



3.4 Demographic distribution

Not all participants chose to complete the few demographic details requested on the reply slip accompanying the information sheet sent out with the invitation. The following details concerning demographic distribution are derived from the information volunteered from the 126 participants.

Table 3.1 Demographic distribution of participants

Subgroup	Male	Age (years): Mean (range)	Time since diagnosed/ duration of caregiving or professional experience with HD (years): range	Number of patients seen in past two years by professionals	Relationship to care receiver with HD/ Profession
People with HD (n=39)	31%	50.5 (27-66)	<1 to 8*	N/A	N/A
Caregivers (n=48)	38%	60.2 (39-80)	1- 20+*	N/A	Relationship information from 45 caregivers (I am a …. to the person I care for). Spouse/ partner 23(51 %) Sibling: 5(11%) Parent 6(13%) Son/ daughter 5(11%) Uncle/ Aunt 3(7%) Granparent 1(2%) Grandchild 1(2%) Step parent 1(2%)
Health Care Professionals (n=39) (nursing= 58%; Medical= 18%	31%	No data	<1-25 (mean = 9.4).	1-200 (mean = 27).	Community Nurse: 6(15%) Pract. nurse: 1(3%) Mental health Nurse: 5(13%) Hosp/ n.home Adult field N: 11(28%) Ph. Therapist: 3(8%) Occ. therapist: 2(5%)

Allied Health Profs= 19% Social Work= 5%)					Spch & Language Therapist: 3(8%) Soc. Worker: 2(5%) Neurologist: 2(5%) Psychiatrist: 1(3%) Clin Psychol: 2(5%) Other medical: 2(5%)

**Mean not calculated due to some vague responses -e.g. 20+ years- Carers and pwHD found estimating duration difficult due to subtle early signs and insidious progression.*

Table 3.2 below provides a summary of the numbers of participants taking part in each of the 3 activities- brainstorming, prioritizing and clustering. In total, 126 participants took part in at least one aspect of the study: 39 people with HD; 48 caregivers; and 39 health professionals.

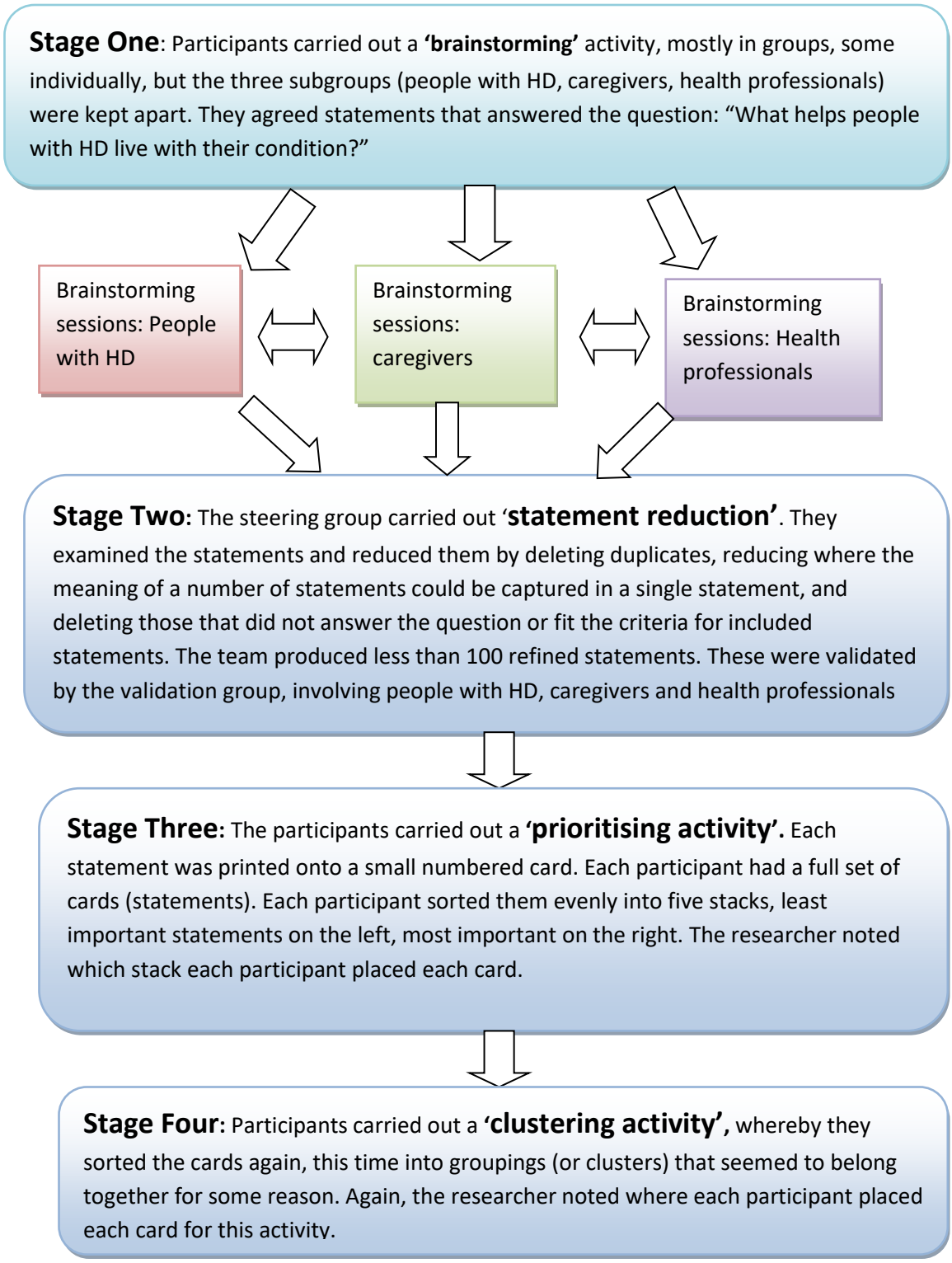
Table 3.2: Summary of Participant involvement in the 3 activities

	Brainstorming	Prioritising	Clustering	Total	Did both prioritising and clustering tasks	Did All three activities
PwHD	29	19	9	39	9	6
Carers	31	26	20	48	9	9
HPs	30	20	20	39	11	11
Total	90	65	49	126	29	26

3.5 Data Collection – detailed explanation.

The concept mapping method involves four stages of data collection, as shown in fig 3.4, and explained in more detail below.

Fig 3.4 Data collection flow chart.



Brainstorming sessions were carried out between October 2009 and August 2010. Clustering and prioritising sessions were carried out between September 2010 and June 2011.

People with HD, caregivers and health professionals took part in the 'brainstorming' sessions separately. The sessions were carried out at locations in eight geographic areas of England, spread between the mid-South Coast to North Lancashire, and the Essex Coast to the East, (Table 3.3).

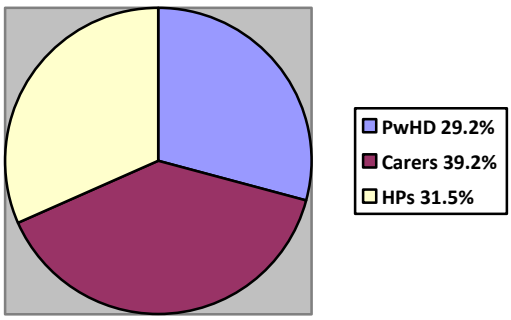
Table 3.3: Numbers of participants at brainstorming sessions by geographic location.

Area	PWHD	Carers	Professionals	Total
Centre 1	2	2	12	16
Centre 2	6	8		14
Centre 3	5	6	7	18
Centre 4	6	5		11
Centre 5			7	7
Centre 6	4	5		9
Centre 7			4	4
Centre 8	6	5		11
Total	29	31	30	90

The largest (and mean) group sizes were: people with HD, n=6, (1.8); caregivers, n=8, (3.4); professionals, n=8, (3.0). The five people with advanced HD were seen individually, to encounter problems in communicating associated with the condition, which would inevitably make group participation difficult. Otherwise two people with HD, one caregiver and six health professionals took part individually, and the remainder took part within groups.

Participants' answers to the question '*What helps people with HD live with their condition?'* were written onto a flipchart by the principal investigator or an assistant (member of the steering group). Comments made by the participants to explain or illustrate the statements were noted.

Fig 3.5 Proportion of total statements (1358 prior to the reduction process) contributed by each subgroup (people with HD; caregivers and health professionals)



The steering group carried out the statement reduction process in which they analysed the statements, and eliminated duplicates where more than one statement had essentially the same meaning, and also any that did not fit the necessary criteria. Where it was decided that a number of statements could be reduced to a single statement that captured their essence, this was done with the intention of capturing what all of the 'legitimate' statements (those meeting the criteria) had been intended to convey, within 100 statements or less. Kane and Trochim (2007) suggest a

reduction to less than 100 statements by these methods if this is possible while ensuring all points that participants intend to make are captured. Their rationale is that the following tasks (prioritising and clustering) may be too onerous for participants if there are more than 100 statements.

The validation group were in general agreement with the reduction process undertaken by the steering group but initiated two changes. The steering group had reduced a number of separate statements about treatment for chorea, to one single statement:

What helps people with Huntington's disease live with their condition is…Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.

The validation group were happy to accept the grouping of most of these activities, but wished to keep separate the statement:

What helps people with Huntington's disease live with their condition is…Medication against chorea.

They felt that this statement was a different issue; separating treatment to reduce chorea as an end in itself from treating chorea to enable a range of activities.

There was divided opinion among the steering group about whether smoking could be grouped together with having alcohol and a range of other social activities said to help people live with HD. The validation group settled this and opted for smoking, and having alcohol to be addressed in two distinct statements, separate from other social activities.

The final reduction exercise resulted in 94 statements. While further reduction would make the sorting sessions more manageable for participants, it was unanimously

agreed that the consequence of doing so would be to sacrifice a point that participants had intended to raise, or combine statements that raised separate issues, which would be unacceptable.

Each of the 94 statements was allocated a randomly allocated number (1-94) and was then printed onto a separate card in preparation for the prioritising and clustering exercises (Fig 3.6).

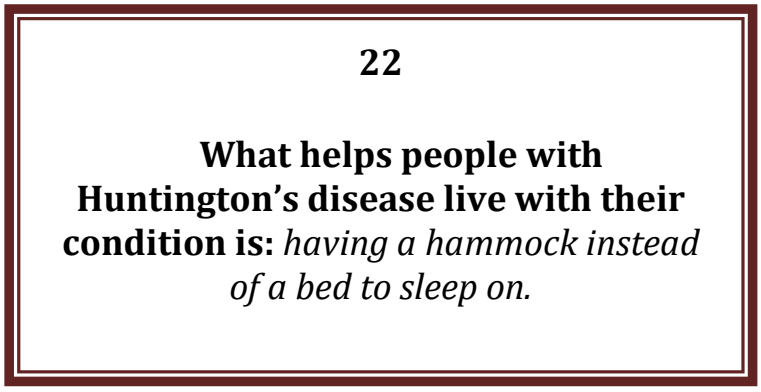


Fig 3.6 Example of a statement card (not an actual statement produced in the study)

In the prioritising task, each participant was given a pack of 94 cards, shuffled to ensure variation in the order in which each statement was viewed. Participants ranked the statements in ascending order of importance by arranging the cards into five stacks. The relatively least important were placed in the first stack, furthest left (stack one) and the most important (in the participant's opinion) were placed in the furthest right stack (stack five). Participants were advised that each of the five stacks of cards should contain at least 17 and a maximum of 21 statements (to avoid people rating nearly all statements as either most important or least important).

After a 15 minute break the second task was to group the cards into stacks, so that all the statements in a particular stack seemed in one way or another, to belong together,

that is, had to do with the same aspect. These groups of statements were called ‘clusters’.

Participants were told that clustering can be done in any way that the individual thought sensible as long as the clusters comprise statements that according to the participant somehow belong together. The fact that there is no ‘right’ or ‘wrong’ way to do this was emphasised, the participants were advised to avoid having a ‘miscelaneous’ cluster in which statements felt to be unrelated might be put together. even if one single statement was felt not to belong with any others it should be placed in its own ‘stack’ to identify it as having a separate theme.

Participants then completed data sheets (appendix 3.3) identifying how individuals had prioritised each statement and which statements had been grouped together to form clusters, and suggesting a name for each cluster, that could best capture the theme of the statements within it.

3.6 Data Entry

The data were entered onto computer using the Ariadne® programme (NcGv/Talcott, Utrecht, The Netherlands, 1995; Severens, 1995). A code for each participant who undertook the prioritising and / or the clustering activities was loaded into the programme by the primary investigator, as was the information from data sheets depicting how each individual prioritised and clustered each statement. Where more than one item shared a mean score, highest priority rating was given to the score with the narrowest standard deviation.

The coding system only identied whether the participant was a person with HD, a caregiver or a health professional. That is, a randomly selected person with HD was

coded as HD1, another as HD2 etc; a randomly selected caregiver was coded as C1, and a randomly selected health professional was coded as P1. To protect confidentiality, only the principle investigator kept access to documents linking participants to codes. Participants who only undertook the brainstorming sessions were not entered into the Ariadne® programme because those data were not needed for that part of the analysis.

As a way of checking the accuracy of the data entry process, another steering group member (HF) randomly selected 10% of participant code numbers, and also entered the data into a copy of the Ariadne® programme. We then compared the entries in both versions to check that they matched. Both sets of entires were found to be accurate and similar.

It is acknowledged that double entry (whereby two researchers each load all data into the programme, and the programme identifies any discrepancy between the two) would have been more ideal. The Ariadne® programme does not accommodate this, and the method we used to check was considered by the steering group to be acceptable as a method of verification of the process.

3.7 Data analysis

The purpose of data analysis was to capture the themes identified collectively by participants, that is, how their generated answers to *'what helps people with HD live with their condition?*' had been conceptualised; and to determine the perceived order of importance for those themes. Analysis was also aimed at identifying any similarities or differences between PwHD, Carer and HP perceptions.

The Ariadne® software uses multidimensional scaling (MDS) and cluster analysis to depict a graphic portrayal of 'distances' between items (statements) by plotting their position on a two-axis matrix, resulting in production of graphic conceptual maps. This approach in health related research using Ariadne® has been adopted by a number of researchers (e.g. de Kok et al 2007; Varekamp et al 2005; Roeg et al 2005).

Strictly, analysis should treat the rating of items as ordinal. The activity of placing cards into five stacks according to importance is in essence the same as using a Likert scale. While stacks 1-5 indicate an increasing level of importance to the participants, we cannot assume that the difference say, between stacks 2 & 3 is the same as the distance between stacks 4 & 5. Jamieson (2004) stresses risks inherent in mishandling data that is ordinal by attributing a scale quality. Jamieson (2004) maintains that calculation of mean scores is appropriate to scale data, whereas median scores are more suited to ordinal data. However, Norman (2010), while acknowledging that this is strictly true, refutes Jamieson's implication that therefore it is inappropriate to use parametric tests to analyse data derived from a Likert scale. Norman emphasises the robustness of parametric methods. He demonstrated that results from a survey using a 10-point Likert scale, analysed as though the data were interval, correlated extremely closely with results after reducing the Likert instrument to 4-point, by grouping together some of the original points so that 0 = 1; 1 and 2 = 2; 3, 4, and 5 = 3; and 6, 7, 8, 9, and 10 = 4. This ensured that the ordinal data was extremely skewed- the concern for Jamieson (2004), being that because distances between points may vary, results may not be accurate. Norman holds that critics who raise this point fail to suggest *how* inaccurate, and overlook the

robustness of parametric methods in their ability to accommodate 'problems' such as low participant numbers, non-normal distribution, and as discussed here, ordinal data.

It seems justifiable in the light of the above debate to calculate means and standard deviations in this current study given that the results will be far more informative than if median scores were calculated. For example, where the median item score for people with HD and for health professionals might be 3, a mean score might be 3.2 for one of the subgroups and 3.6 for another, demonstrating that there is a calculable difference in the item's importance between the subgroups.

Developers of specialised concept mapping programmes (Kane and Trochim, 2007; Severens 1995) clearly adopt this view, as both Ariadne® and the 'Concept Systems' software (www.conceptsystems.com) do calculate mean scores and standard deviations. The practice is well established, with many examples of published research in health based on calculations obtained through the use of either of these two concept mapping programmes (e.g. Nijman et al 2011; Anderson et al 2011; Scahill et al 2010: van Bon-Martens et al 2011).

Norman (2010) suggests that the widely adopted stance that parametric tests are based on assumptions of normality tends to be misunderstood. The means, not the data have to be normally distributed and he points out that the Central Limit Theorem demonstrates that for sample sizes of more than five or ten per group, the means are 'approximately normally distributed regardless of original distribution'.

Nevertheless, the broadly held view (e.g. Field 2009) would demand that in order to consider whether any differences between subgroups were statistically significant regarding how specific variables were prioritised it was first necessary to establish

whether data were normally distributed to determine whether parametric or non-parametric testing would be more appropriate and this was complied with.

SPSS™ software (SPSS inc. Chicago Illinois) was used to carry out the Kolmogorov-Smirnov and Shapiro-Wilk tests for normality. The latter is suggested as more appropriate for small samples (Field 2009). For both tests the null hypothesis is that the data are normally distributed and so a significance value of <0.05 means that the null hypothesis can be rejected and that the data are therefore not normally distributed.

Appendix 3.4 provides results of the test for normality carried out. The table shows that significance for each subgroup regarding almost all variables (i.e. except for 12.5%, or 24 of 192 values) was <0.05 indicating that data were not normally distributed. Visually on the quartile-quartilie (q-q) plots deviation from the line expected if data were normally distributed appears only slight in nearly all cases, but in view of the small numbers of participants in this study, it was considered appropriate to rely only on numeric results for the test for normality and therefore reporting is based on non-parametric tests.

Reported statistics comparing scores between subgroups, unless otherwise stated were derived from the Kruskall-Wallis test, a non-parametric equivalent of the one-way ANOVA. However, in line with suggestions by Norman (2010) the one-way ANOVA test was also run and produced similar results.

It is possible to calculate confidence intervals from mean scores and standard deviations but this is felt to be unjustified as it would not add value amd so was not done**.** The concept mapping method is developed to calculate mean priority scores and the position of statements on an NxN matrix on the basis of frequency with

which a given statement was placed in a cluster stack along with another given statement. Reliance on mean scores is widely accepted and consensus among researchers who employ the method seems to be against reporting confidence intervals (e.g. Roeg et al, 2005; de Kok et al 2007; Nijmen et al, 2011).

Selecting subgroups for analysis

The study aimed to generate statements about what helps people live with HD from a broad range of perspectives and obtain an overall inclusive priority order and grouping of statements. The concept mapping method is designed for this, but can also facilitate comparison of subgroups. There is no justifiable rationale for comparing data from groups separated according to gender or age. Each group that generated statements included both genders (though as shown male representation is relatively low) and an age range encompassing mid to late life in keeping with the general pattern of HD duration.

However, people with HD, family caregivers and health professionals generated statements separately as subgroups at the outset and as these are different 'roles' with regard to living with HD, comparing and contrasting the three perspectives was considered to be legitimate. Ho et al (2006) suggest that comparison of perspectives between patients with HD and those of carers can provide valuable insight into management approaches most likely to succeed. Understanding of similarities and differences between patient, carer and professional perspectives more generally can help to improve the working relationship and clarify needs (Walters et al 2000).

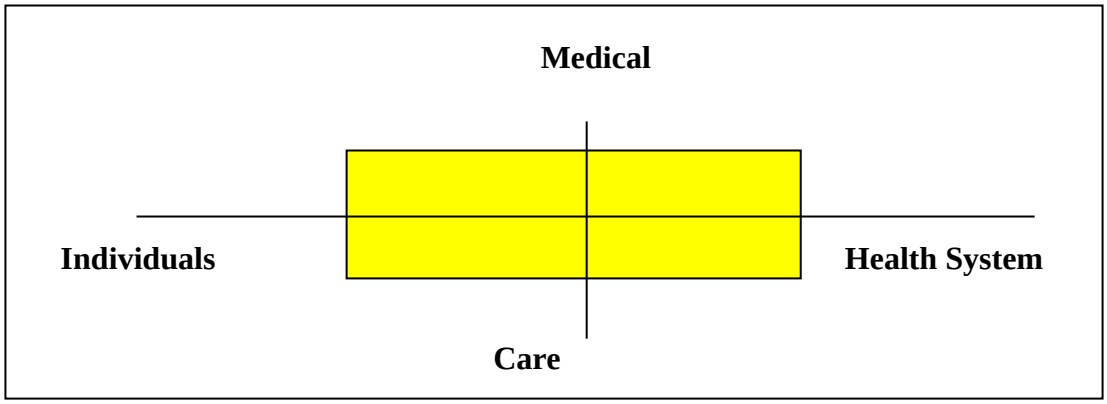
Living with and providing for people with HD in the context of a nursing home during the advanced stages of disease, where a paid team provide 24 hour care in facilities dedicated to that purpose must intuitively be different to managing at home

in the community where family members gradually assume a role as disease symptoms insidiously develop. This perceived difference is the justification for drawing a distinction between the nursing home and community contexts. For these reasons analysis was undertaken for patient, carer, health professional, community-based and nursing home-based subgroups.

Cluster Analysis

The Ariadne® programme forms a visual 2 dimensional matrix and plots the statements according to 'distances' representing the frequency with which statements had been grouped together by participants. The graphic rectangular representation produced is divided into four equal quadrants by two axes, and allows the researcher to name the axes. Each end of each axis was given a name intended to sum up the content of statements situated near to that area of the concept map (see Fig 3.7). The steering group verified the name given to each end of each axis on the concept map.

Fig 3.7: Matrix with labelled axes



To clarify, it was decided that the statements the software plotted near to the top centre of the rectangle tended to relate to medical input. Those statements positioned near the far right tended to relate to the health care system whereas those at the far

left were mostly to do with living as an individual. Statements that Araidne® located on the lower part of the rectangle tended to relate to some aspect of care.

The number of clusters that group the statements meaningfully tends to be a minimum of eight and a maximum of 20 (Jackson and Trochim 2002). Using the Ariadne® programme and beginning with six clusters the number of clusters was increased until, conceptually, this seemed to make sense, in that natural groupings of the statements, according to their location on the matrix, seemed to be optimally captured by nine clusters. At this point, reducing the number of clusters by one seemed to fail to acknowledge distance between some items from others on the matrix that are forced into the same cluster. Increasing the number of clusters by one separated out items that, according to their position on the matrix, seemed to belong with items in a neighbouring cluster. The process was checked and verified by the steering group and the validation group and all agreed that a nine-cluster solution should be adopted. The results section shows that when the data for subgroups (e.g. people with HD, caregivers, health professionals, and also participants with HD experience in a nursing home context and in a community-based context) were analysed a different number of clusters was agreed (in most cases a ten-cluster solution). In each case the number of clusters was agreed through the process described above.

The element of subjectivity in this process is acknowledged, and is considered to be appropriate in this research method (Jackson and Trochim 2002).

Latent preferences

The Ariadne® programme is also able to present maps that plot a relationship between individual participants and the matrix. This also uses multi-dimensional

scaling. For example, a participant who frequently prioritises statements that relate to care, will be plotted near to the lower end of the vertical axis on the map (the care-medical axis). The participant would be considered to have a 'latent preference' towards prioritising statements concerned with care.

3.8 Summary

In the absence of good quality evidence for the effectiveness of interventions for HD, it is valuable towards planning provision of services, care, support and treatments for people affected by the disorder to understand what helps to live with the condition from the perspectives of key stakeholders- patients, family caregivers and health professionals. This study aims to address this need by generating a graphic, conceptual map.

Various qualitative approaches to determine what is most helpful towards living with HD were considered but the inclusivity of ideas, the generation of concepts or themes based on groupings of specific statements and the quantified relative importance of those themes accommodated by the concept mapping method in addition to the resulting visual conceptual map were factors that contributed to choosing the concept mapping method.

Chapter Four

Results

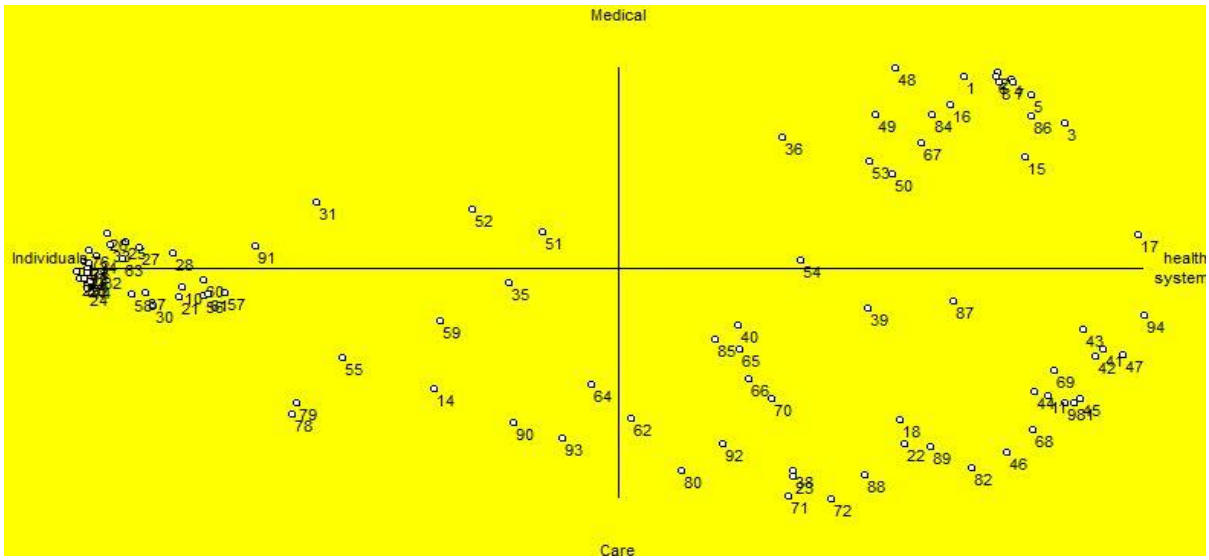
The study aims were first to identify factors that help people with HD live with their condition from the combined perspective of people with HD, caregivers and health professionals, and second to generate a graphic map illustrating how these factors are conceptualised by the participants, using concept mapping. The third aim was to establish a perceived order of importance for conceptual clusters of factors that help and the fourth was to compare and contrast perspectives of participant subgroups.

In order to meet these aims I collected statements generated in brainstorming sessions in which participants agreed ways to complete the semtence *'What helps people with HD live with their condition is…'.* Participants then prioritised the statements by allocating to each a score of 1-5 (1=least important, 5= most important) and grouped the statements according to their similarity (clustering).

To avoid unnecessary repetition results regarding numbers of participants recruited and their characteristics were reported in the previous 'methods' chapter. This chapter presents the results from the prioritising and clustering exercises.

The Ariadne® software uses multidimensional scaling to represent the data graphically on concept maps. Fig 4.1 is a point map that depicts the way the participants sorted the 94 statements. Each numbered point represents a single statement and the distance between statements on the map indicates the frequency that statements were grouped together in clusters. Statements placed together into the same cluster by many of the participants appear close together on the map whereas statements less often grouped into the same cluster are further apart. The statements that each numbered data point on the map relates to are listed in appendix 4.1.

Fig 4.1[a] Concept map for all participants depicting statement locations in relation to matrix



In Fig 4.1[a] each numbered point on the map depicts the positioning of individual statements in relation to each other and to the two axes (individuals – health care system; and medical – care). That is, statements that participants often grouped together in cluster stacks are represented close together. The statements and corresponding numbers are listed in appendix 4.1.

4.1 Clusters

Analysis of the clustering of the 94 statements using Ariadne® software resulted in the formation of nine clusters for 65 participants representing the three stakeholder subgroups (people with HD, Carers and professionals). The process by which a nine-cluster solution was arrived at is explained in the methods chapter. Table 4.1 presents the clusters in priority order (cluster 1= most, cluster 9= least important). A high mean score indicates that on average statements within a cluster had been prioritised highly by participants. The maximum possible score would be five, which would have indicated that all participants had given all statements within the cluster highest priority; the lowest possible mean score would be one (neither of these extremes occurred). The table shows the number of statements within each cluster. The highest scoring cluster contained only one statement: *'what helps people with*

HD live with their condition is if there is expert assessment and treatment of mental health problems (e.g. depression, anxiety) occurring along with HD'.

Fig 4.1[b] Cluster map for all participants

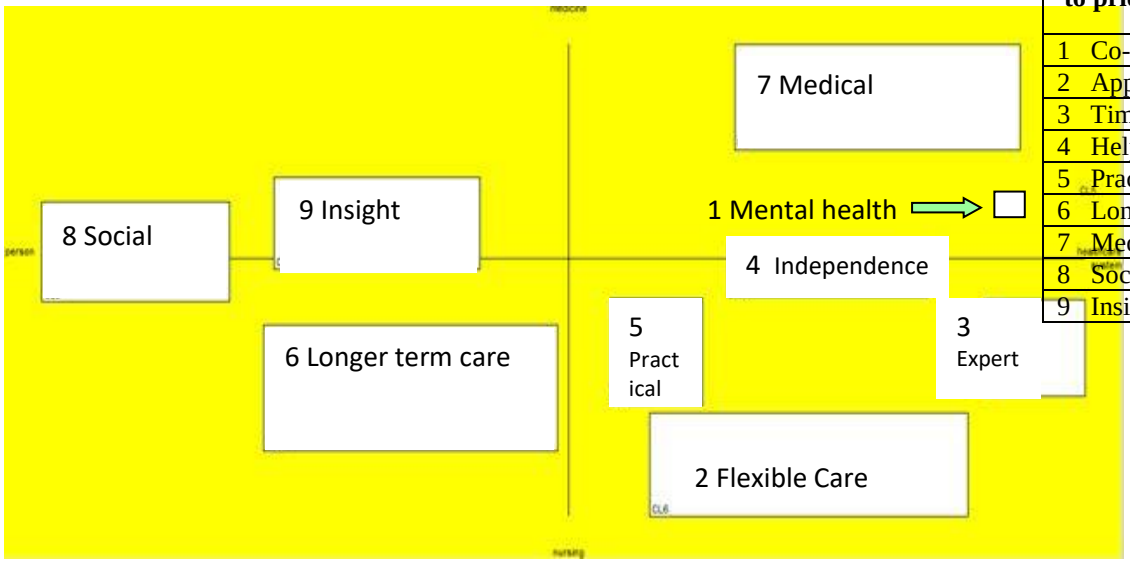


Table 4.1 Cluster table for All Participants

Cluster Name (numbered according to priority order 1= most important)	Mean importance rating	No. of state-ments
1 Co-morbid mental health problems	3.72	1
2 Appropriate flexible care	3.32	7
3 Timely integrated expertise	3.28	17
4 Help to independence	3.15	8
5 Practical engagement	3.15	2
6 Longer term care issues	3.12	8
7 Medical input	2.93	18
8 Social living	2.90	29
9 Insight	2.77	4

The statements that make up each of these nine clusters are identified and discussed below.

Cluster analysis.

Tables 4.1.1 to 4.1.9 below show the statements that make up each cluster in the order of the mean importance rating for all participants (that is, the three subgroups- people with HD, carers and health professionals combined).

Cluster 1, Co-morbid mental health problems

The identification of assessment and treatment for mental health issues as the highest priority among factors that help in living with HD as rated by the combined subgroups (people with HD, carers and health professionals) is the key finding of this study. The cluster contains only one singles statement. Mental health problems were discussed in every brainstorming session and in the reduction process the statement was formed to capture the essence of many original statements about living with a variety of co-morbid mental health conditions.

Table 4.1.1 Content of cluster 1, Co-morbid mental health problems.

Cluster 1 **Statement**	**Mean importance rating**
If there is expert assessment and treatment of mental health problems (e.g.depression, anxiety) occurring along with HD	3.72

Cluster 2, Appropriate flexible care

The second highest priority cluster referred to the importance of 'appropriate flexible care'. One tale told by a caregiver during a brain storming session seemed to typify the perceived problems that underpin the seven statements within the cluster: *'She* [paid Health Care Assistant] *came to the house one morning and said she'd come to help wash and dress my husband. I told her it took me years to get the routine right so he doesn't go into a tantrum. If you do up the shirt buttons top down instead of bottom upwards, or shave him the wrong way, he'll become aggressive. So I asked*

her to help get the children ready for school, to free me to help him. She took that to mean I refused the help she'd been sent to offer. No help's better than inflexibility'.

Being offered care that is regarded by recipients as inappropriate or inflexible seems to be the cause of much angst in families affected by HD.

Table 4.1.2 Content of cluster 2, Appropriate flexible care

Cluster 2 **Statements**	**Mean importance rating**
If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.	3.86
If care is available where the person with HD needs it.	3.75
Not being cared for in an inappropriate setting (e.g. psychiatric or elderly setting where staff don't know about HD).	3.72
If informal carers have training in HD	3.17
If a care home environment is made to feel as home should feel	3.02
If visiting carers are flexible regarding the specific type of care they provide	2.98
If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).	2.71

Cluster 3, Timely integrated expertise

Expertise is clearly a highly important issue for participants. Having access to professionals who are expert in HD was the highest scoring individual statement overall and in brainstorming sessions it was noted that dealing with GPs, nurses and neurologists who do not appear to understand the condition well can cause families enormous frustration. Those who had been referred to a national specialist HD centre were considered highly fortunate and invariably commented that living with HD became very much more manageable following the first visit. Many participants commented that they would be prepared to travel to access integrated interdisciplinary specialist expertise. Timeliness of assessment, interventions and receipt of equipment was seen as a natural benefit of access to experts who know HD, and know the family well and can plan ahead as opposed to responding to

crises. This explains the importance of continuity of staff, also highlighted within cluster three.

Table 4.1.3 Content of cluster 3, Timely integrated expertise.

Cluster 3 **Statements**	**Mean importance rating**
Access to professionals who are expert in HD	4.12
If care is available when the person with HD needs it	3.92
Access to a multi-disciplinary team that is expert in HD	3.82
Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation	3.80
Being referred to a specialist National HD centre	3.52
Having continuity of care staff, (e.g., professional carers in homes or that visit at home).	3.52
Regular and timely access to health and social care assessment	3.46
When service provision is planned for when it will be needed, rather than thought about once things have already got difficult	3.43
Continuity of health and social care professionals	3.31
If there is information about HD for health professionals in general medical settings	3.11
If there is information about HD for health professionals in general medical settings	3.11
Effective multi-disciplinary working	3.09
If professionals help the person with HD to get a life	2.91
Effective interagency working	2.83
If professionals follow up patients who do not keep appointments	2.63
If people with HD know the role of each professional	2.63
Timeliness in access to care homes	2.54

Cluster 4, Help to independence

The central themes for cluster three of timeliness and expertise also feature in cluster four in the context of having help towards independence. Aids and adaptations were seen as important so long as they are provided at the right time (when needed) and with regular assessment to ensure continued suitability. For people with HD for whom tube-feeding is regarded as helpful the introduction of this intervention needs to be timely and participants commented that this would avoid a crisis-led approach in which unhelpful decisions can be made hurriedly. A timely approach can empower people with HD to maximise independence by being enabled to make choices about this issue (Simpson 2007). The involvement of speech and language therapists was a factor associated with independence not only for their part in helping to overcome swallowing problems but also for help with communicating.

Early assessment and a responsive approach to changes in ability to initiate and take part in communication throughout the various stages of HD progression are called for in recently formulated European guidance concerning speech and language therapy for people with HD and their families (Hamilton et al 2012). The guidance recognises that communicating problems associated with HD challenge families, not only individuals.

The importance of having the right level of information about HD at the right time was stressed within this cluster. This was often seen as provided by family support workers from the HDA. Another aspect of choice grouped into the cluster concerned with help to independence was that of deciding whether to see a male or a female GP.

Cluster four presents an important theme for professionals to bear in mind.The loss of independence is associated with a higher risk of suicide among people with HD (Novak and Tabrizi 2010). Independence can be seen as integral to the aims of rehabilitation generally and specifically for people with HD (Ciancarelli 2013).

Table 4.1.4 Content of cluster 4, Help to independence

Cluster 4 **Statements**	**Mean importance rating**
Speech and language therapy involvement to help with swallowing problems	3.92
Speech and language therapy involvement in maintaining ability to communicate	3.62
Having support from the Huntington's Disease Association.	3.58
Having timely access to appropriate aids and adaptations	3.28
Having access to the right level of information about HD at the right time	3.22
Regular timely assessment for aids and adaptations	3.18
Timely introduction of tube feeding	2.55
Having a choice about seeing a male or a female GP	1.85

Cluster 5, Practical support

There was considerable discussion within the steering and validation groups about why the analysis process had led to the two statements in table 4.1.5 (below) being

grouped together and apart from other clusters. It was generally agreed that this had occurred because of a tendency among the participants to avoid grouping 'having support from the HDA' with 'being involved in a HDA support group', and also avoid grouping 'advice on foods and textures' with issues to do with 'swallowing' in cluster four. Active involvement in a local branch of the HDA was evidently seen as a separate and much more practical issue than being a member of the national organisation. Rosenblatt et al (1999) suggest that people with HD are often reported by family caregivers as disengaged. According to Rosenblatt et al (2009) this is largely due to lack of internal initiation and can be countered by external initiation which can re-engage the person with HD in work and social activities. It seems reasonable that membership of a national organisation may not necessarily stimulate drive whereas involvement in a local branch might. This appears to be the point identified by participants in this study.

Although speech and language therapy can address problems with communicating and with swallowing, it was felt that having practical advice on textures and kinds of food to eat may have seemed to participants to go beyond problem-solving and to be a more proactive way of being supported to live, in common with having an active role within a support group. Indeed, Aubeeluck et al (2011) cite the importance of food being of appropriate size and texture as an example not only of safeguarding against or reducing dysphagia but as a means of improving quality of life.

Table 4.1.5 Content of cluster 5, Practical support.

Cluster 5 **Statements**	**Mean importance rating**
Being involved in a Huntington's Disease Association support group	3.48
Practical advice on specific foods and textures	2.83

Cluster 6, Longer term care issues

In cluster six, eight statements brought together issues such as enabling daily living through the provision of bathroom aids and aids for communication and falls-prevention; having training to use the aids and being able to make choices about ongoing and end of life care. Aspects of life in institutional long term care such as being able to go out and engage in the community while a nursing home resident and having meaningful relationships with other nursing home residents were included in this cluster as were concerns about family caregivers: having a break from caring and being listened to and believed. In a care of the elderly context Wolff (2013) found that satisfaction with life can be enhanced and depression reduced more effectively among nursing home residents by activities such as frequent in-house social events aimed at fostering friendships between fellow residents than by visits from family which are of course also positively influential.

Anderson and Dabelko-Schoeny (2010) report emotional and physical benefits for civic engagement among older adults but found that opportunities to do so once admitted to a nursing home are limited. Social and practical barriers (for example ageism and transportation) are identified as reasons for this and the authors call for social work action to overcome these obstacles. This study supports a need for such action on behalf of a younger cohort also: nursing home residents with HD.

Table 4.1.6 Content of cluster 6, Longer term care issues

Cluster 6 **Statements**	**Mean importance rating**
If the family carer has a break from looking after him or her.	3.80
Having aids to enable daily living (eg. aids that help with communication, mobility, furniture, bathroom and falls prevention).	3.63
Making choices about care including end of life care in advance and having the choice respected	3.59
If carers are listened to as an adult and believed and respected	3.49
Having assistance to eat and drink	3.35

Being able to go away from the care home to engage in community activities	2.78
Having appropriate training to use equipment	2.35
Having meaningful relationships with other care home residents	2.00

Cluster 7, Medical input

Within this cluster partipants highlighted that medication can be helpful in reducing chorea, particularly if doing so is aimed at improving ability to carry out activities such as sleeping and having a sex life and to reduce stress and fatigue. In a separate statement the value of medication to help sleep was reiterated. Timely reviews of medication were called for and having the goal of a normal life when prescribing medicines was situated in this cluster. Participants cautioned against prescribing sedative medication for the sole purpose of managing behaviour and stressed that the effects of medication vary from patient to patient and that side effects can be seriously detrimental to living with HD. Assessment for aggressive behaviour was seen as helpful and recognition that behavioural problems and impulsivity are serious aspects of HD was called for. Availability of genetic testing and pre-implantation diagnosis were viewed as part of medical input.

A wish to be able to try medicines experimentally as individuals, without waiting to find out if there will be a trial was expressed within the cluster.

Table 4.1.7 Content of cluster 7, Medical input

Cluster 7 **Statements**	**Mean importance rating**
Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.	3.75
Medication against chorea	3.48
A prompt diagnosis	3.32
Having appropriately timed reviews of medication	3.22
Having medicines to help sleep	3.22
Having the goal of a normal life when considering the use of medication	3.13
Pre-test and post-test counselling	3.11
Assessment of management of aggression if it occurs	3.06
The availability of a genetic test	3.03
If obsessive behaviour and impulsivity are recognised as serious symptoms of HD	3.02

Recognition that antipsychotic medication can be helpful in HD	2.85
Having the opportunity to be involved in research	2.83
Availability of pre-implantation diagnosis	2.62
Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).	2.57
Knowing that sedative medication will not be given solely to manage behaviour	2.51
Knowing that side effects of drugs for HD can be serious	2.45
Knowing that the effects of medications for HD are variable	2.42
Recognition that facial and neck pain can be a problem in HD.	2.13

Cluster 8, Social living

This was the largest cluster in terms of the number of included statements. The importances of supportive family and friend networks and of a stable partner relationship were expressed as was being able to talk freely about HD in the home situation. Being able to drive, smoking cigarettes and drinking alcohol were grouped among the statements concerned with social living. Other factors included in this cluster were having hobbies and pastimes, having meaningful conversations, maintaining a positive outlook, being able to get out and about, accessible transport facilities and having strategies to enable eating out without embarrassment. People with HD were felt to need to be able to engage in intellectual as well as physical activities and there was a call for employers to make reasonable adjustments to enable continued working after diagnosis. Contrasting approaches to money management are suggested as helpful.

Table 4.1.8 Content of cluster 8, Social living

Item	**Mean importance rating**
Having a supportive network of family and friends	4.09
Having a familiar daily routine.	3.95
Having family and friends who offer practical support	3.88
Having a stable relationship with a partner	3.82
If the person with HD has help to be able to get out and about	3.62
Having a positive outlook on life	3.49
If he or she is able to talk freely and openly about HD within the family	3.57
Having opportunities for meaningful conversation	3.49
Having help to socialise	3.35
If there is availability of food that is appetising and manageable	3.17
Living for today	3.08
Participating in physical activities	3.05

Having someone to sort out his or her access to financial benefits	3.03
Knowing in advance the detailed plans for what will happen today and / or at forthcoming events	2.83
Being able to take up hobbies and pastimes	2.83
Having practical strategies to enable him or her to eat out without embarrassment	2.78
Having community understanding	2.77
Having accessible town, transport and shop facilities	2.71
If employers make reasonable adjustments to enable continued employment	2.66
Direct access to cash benefits to spend on what is needed now	2.63
Participating in intellectual activities	2.60
Being able to undertake domestic activities such as cooking, gardening and cleaning	2.55
If they are enabled to maintain the roles that they feel suit their gender	2.51
Still being able to drive.	2.28
Spending money while he or she is able to	2.12
Having alcohol	1.89
Having cigarettes	1.83
Being prudent with money	1.68
Having a smoking aid	1.66

Cluster 9, Insight

The issue of the extent to which a person with HD might posess insight is the subject of further tension between perspectives that is also discussed below with regard to subgroup differences. This cluster contained four statements to do with the person with HD being able to accept his or her diagnosis, and being listened to and believed; and also knowing and not knowing if family members have a positive HD diagnosis. Insight proved to be a controversial issue. All participants appeared to see value in knowing the genetic status of family members regarding HD but some also placed some value on not knowing. One parent described the one consolation felt by him and his wife after their 20+ year-old son died in an accident. They shared a sense of relief that they had kept the family legacy of HD a secret from their son and that he had been able to enjoy living as long as he did without ever having been troubled with the knowledge of his at-risk status.

Table 4.1.9 Content of cluster 9, Insight

Item	Mean
If they are listened to as an adult and believed and respected.	3.72
If they accept the diagnosis.	3.26
Knowing if other family members are genetically positive for HD	2.23
Not knowing whether other family members are genetically positive for HD	1.86

Comparison between perspectives of the three subgroups

Some differences in how the statements were prioritised and clustered were found when the data from subgroups (people with HD, caregivers, health professionals, participants with HD experience from a nursing home context / community-based context) were analysed separately and compared.

People with HD

The priority order of the 94 statements for this subgroup is shown in Appendix 4.3. A ten-cluster solution was arrived at following the process and Table 4.2 below demonstrates that the highest scoring cluster for people with HD was 'Being Trusted'. Appendix 4.3 shows the content of each of the clusters for this subgroup.

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Fig 4.2 Cluster map for People with HD

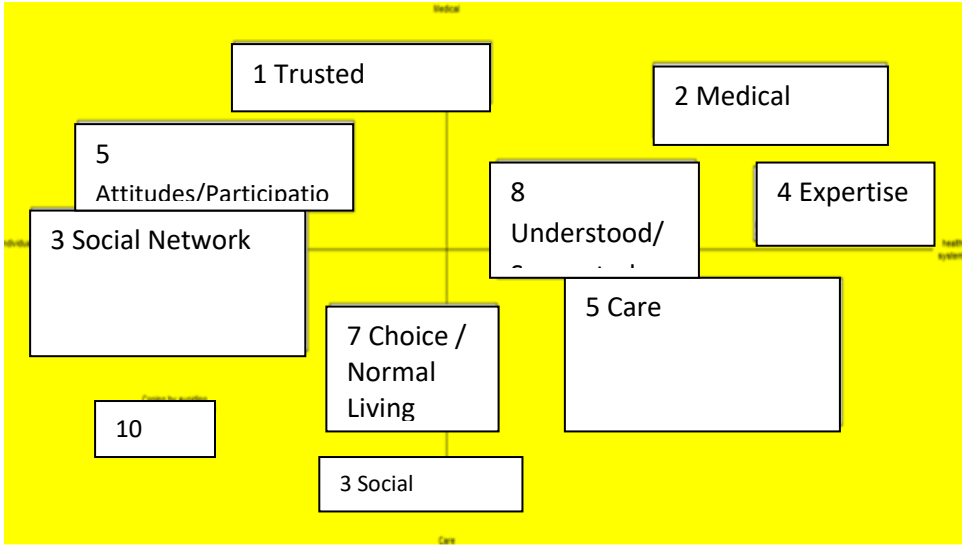


Table 4.2: Clusters for people with HD in priority order based on mean score (Cluster 1 = most important).

Rank order and cluster name (1=most important)	Mean importance rating
1 Being trusted to take part	3.29
2 Medical input	3.27
3 Social network	3.22
4 Expertise	3.19
5 Attitudes and participation	3.13
6 Care	2.95
7 Choice & help to continue normal living	2.94
8 Being understood and supported	2.94
9 Care homes	2.29
10 Coping by avoiding	1.84

The highest scoring cluster contains five statements including *'what helps people with HD live with their condition is if they are listened to as an adult and believed and respected' and '…still being able to drive'* as shown in Table 4.2.1 below. Tables 4.2.1 to 4.2.10 show the statements that make up each cluster in the cluster map for people with HD.

Cluster 1 for people with HD: being trusted to take part.

For people with HD the cluster rated as most important emphasises their wish to be trusted to say what the need and don't need and determine what they are able to do. During the generation of the statements in this cluster in brainstorming sessions some people with HD expressed resentment about family members and professionals reaching conclusions about whether they should still be able to the shops or continue driving. The participants were mostly support group members and valued involvement in the groups, especially when this enabled them to engage in activities they would not manage without the group such as gliding, absailing and camping. Participation in research gave some a feeling that despite their own deterioration, they can contribute to preventing their children from sharing the same fate.

Table 4.2.1, Cluster 1, Being trusted to take part (PwHD) **Statements (1= most important).**	**Mean importance rating**
If they are listened to as an adult and believed and respected.	3.79
Still being able to drive.	3.42
Being involved in a Huntington's Disease Association support group	3.32
Having the opportunity to be involved in research	3.21
Having accessible town, transport and shop facilities	2.74

Cluster 2 for people with HD, Medical input.

This cluster of statements mostlty concerned with interventions such as medical treatments, genetic testing and counselling was rated highly by people with HD, with a score very close to that of cluster 1 (mean rating =3.27 compared to 3.29). In brainstorming sessions people with HD were frequently noted to express frustration at experiencing symptoms of HD despite going to see consultants. The hope for a cure was mentioned only among this subgroup. Some keenly expressed willingness to try experimental medications and the purpose of treatments was frequently noted as being enabled to continue or return to enjoying 'normal' life activities.

Expectations that medical expertise should be able to improve life seem to be stronger among people with HD than among carers or professionals.

Table 4.2.2 Cluster 2, Medical input (PwHD) **Rank order and name of statements (1= most important).**	**Mean importance rating**
If there is expert assessment and treatment of mental health problems (e.g. depression, anxiety) occurring along with HD	3.79
Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.	3.68
Having support from the Huntington's Disease Association.	3.68
The availability of a genetic test	3.58
Medication against chorea	3.53
If obsessive behaviour and impulsivity are recognised as serious symptoms of HD	3.32
A prompt diagnosis	3.21
Having the goal of a normal life when considering the use of medication	3.17
Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).	3.16
Having access to the right level of information about HD at the right time	2.84
Pre-test and post-test counselling	2.84
Knowing that side effects of drugs for HD can be serious	2.42

Cluster 3 for people with HD, Social network

People with HD often referred to the importance of family and friends, of a stable relationship, familiar routines in the home and other aspects of social living identified in table 4.2.3 during brainstorming sessions. Some voiced the opinion that life with HD would be very much more difficult for people without a strong social network and a limitation to this study acknowledged earlier is failure to recruit many participants in that situation, the overwhelming majority being support group members. One participant said that stability was so important that it over-rode the importance of staying with her husband: '*I told him to think about the future when I'll be as ill with HD as my father was, and decide if you're going to stay the course- if not leave now… I'll cope with you or without you, but not with the disruption of you changing your mind at a later stage and leaving home then. He told me he'd stay, but I said, no, think about it some more and tell me in a month- then you have*

to promise to stick to your decision however difficult my behaviour becomes. He did decide to stay.'

Table 4.2.3 Cluster 3, Social Network (PwHD) **Statements**	**Mean importance rating**
Having a supportive network of family and friends	4.11
Having a stable relationship with a partner	4.00
If the family carer has a break from looking after him or her	3.89
Having help to socialise	3.63
If carers are listened to as an adult and believed and respected	3.63
Having a familiar daily routine.	3.47
Having opportunities for meaningful conversation	3.26
Having assistance to eat and drink	2.89
Being able to undertake domestic activities such as cooking, gardening and cleaning	2.89
Knowing if other family members are genetically positive for HD	2.58
If they are enabled to maintain the roles that they feel suit their gender	2.37
Being prudent with money	1.95

Cluster 4 for people with HD, Expertise

The fourth most important cluster for people with HD is 'Expertise' and the statements within that cluster are listed below in table 4.15. The statements about expertise specific to HD for professionals and teams of professionals scored highest in the cluster.

Table 4.2.4 Cluster 4, Expertise (PwHD) **Statements**	**Mean importance rating**
Access to professionals who are expert in HD	4.32
Access to a multi-disciplinary team that is expert in HD	3.58
If there is information about HD for health professionals in general medical settings	3.58
Being referred to a specialist National HD centre	3.21
Recognition that antipsychotic medication can be helpful in HD	3.05
Having medicines to help sleep	2.89
Having appropriately timed reviews of medication	2.79
Knowing that the effects of medications for HD are variable	2.74
Knowing that sedative medication will not be given solely to manage behaviour	2.58

Cluster 5, Attitudes and participation.

In this cluster the statements (as listed in table 4.2.5) link attitudes such as having a positive outlook and accepting the diagnosis, living for today and talking openly and freely about HD; to having the support and help to participate- this aspect of taking part is different to that highlighted in cluster one where the emphasis was on being trusted to shop and drive. Here, areas where there may be more dependence on others to enable participation are grouped together.

Table 4.2.5 Cluster 5, Attitudes and participation (PwHD) Statements	Mean importance rating
Having a positive outlook on life	4.11
Having family and friends who offer practical support	3.84
If they accept the diagnosis.	3.47
If the person with HD has help to be able to get out and about	3.42
If there is availability of food that is appetising and manageable	3.32
Participating in physical activities	3.26
Being able to take up hobbies and pastimes	3.26
Having someone to sort out his or her access to financial benefits	3.21
Living for today	3.11
Participating in intellectual activities	3.00
Practical advice on specific foods and textures	2.89
If he or she is able to talk freely and openly about HD within the family	2.89
Having practical strategies to enable him or her to eat out without embarrassment	2.84
Spending money while he or she is able to	2.58
Direct access to cash benefits to spend on what is needed now	2.58
Knowing in advance the detailed plans for what will happen today and / or at forthcoming events	2.32

Cluster 6 for people with HD, Care.

'Care' was identified as the theme connecting the 18 statements in the sixth most important cluster for people with HD (Table 4.2.6). As will be demonstrated in later sections of this thesis, care aspects generally held a higher priority among family caregivers and health professionals who tended not to attribute participation and autonomy with as high priority as people with HD did.

Table 4.2.6 Cluster 6, Care (PwHD) Statements	Mean importance rating
Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation	3.67
Not being cared for in an inappropriate setting (e.g. A psychiatric or elderly setting where staff don't know about HD).	3.58
Having continuity of care staff, (e.g., professional carers in homes or that visit at home).	3.58
If care is available when the person with HD needs it.	3.58
Continuity of health and social care professionals	3.47
If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.	3.47
When service provision is planned for when it will be needed, rather than thought about once things have already got difficult	3.32
Regular and timely access to health and social care assessment	3.16
If professionals work with all members of the family	3.05
If a care home environment is made to feel as home should feel	2.79
If people with HD know the role of each professional	2.63
If professionals follow up patients who do not keep appointments	2.53
Regular timely assessment for aids and adaptations	2.47
Effective interagency working	2.42
Having timely access to appropriate aids and adaptations	2.37
Recognition that facial and neck pain can be a problem in HD.	2.37
Timeliness in access to care homes	2.32
Timely introduction of tube feeding	2.26

Cluster 7 for people with HD, Choice & help to continue normal living

A statement about being able to make choices about aspects of care including end of life care tended to be placed by people with HD into the same cluster as the other 10 statements shown in table 4.2.7 most of which are about having help- including training to use equipment- to overcome problems associated with HD such as swallowing and communicating difficulties, and be able to get on with ordinary daily activities and carry on in employment as far as possible. The statement about availability of pre-implantation diagnosis also emphasises the importance of being enabled to make choices. The validation group in agreement with the research team concluded that the theme binding these statements making up the 7th most important cluster for people with HD was being able to continue making choices while being helped to live as normally as possible.

Table 4.2.7 Cluster 7, Choice & help to continue normal living **Statements**	**Mean importance rating**
Speech and language therapy involvement to help with swallowing problems	3.95
Speech and language therapy involvement in maintaining ability to communicate	3.74
Making choices about care including end of life care in advance and having the choice respected	3.58
If visiting carers are flexible regarding the specific type of care they provide	3.28
Having aids to enable daily living (eg. aids that help with communication, mobility, furniture, bathroom and falls prevention).	3.11
If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).	2.95
Availability of pre-implantation diagnosis	2.53
If employers make reasonable adjustments to enable continued employment	2.47
Having alcohol	2.32
Having appropriate training to use equipment	2.26
If professionals help the person with HD to get a life	2.21

Cluster 8, for people with HD, Being understood and supported

The steering and validation groups agreed that the theme running through six statements in this eighth priority cluster for people with HD was the need to feel

understood and supported. Some overlap between some clusters was acknowledged, for example, the issue of choice also appears here, as it does in cluster seven. But it was felt that the main emphasis demonstrated by people with HD in frequently grouping the statements in Table 4. together, and apart from statements in other clusters was focussed on ways in which they could be helped by being understood- for example through training for informal carers, by having aggressive behaviour carefully assessed and by there being greater awareness about HD in the community; and supported by effective multi-disciplinary working and having care provided where needed. Notes taken in brainstorming sessions suggest many people with HD felt care often needed to be given at home but accepted that it can become preferable to have care in a nursing home, and if so this should be near home so that family may visit and day visits back home are manageable. While prepared to travel distances for expertise to assess and treat, and arrange and organise care (see cluster 3 all participants, Table 4.1.3), people with HD did not appreciate receiving care long distances from home. In the context of the statement being set into this cluster, the choice regarding GP gender was taken by the steering and validation groups to mean that this would help facilitate a comfortable, open and supportive dialogue.

Table 4.2.8 Cluster 8, Being understood and supported (PwHD) **Statements**	**Mean**
If care is available where the person with HD needs it.	4.05
Effective multi-disciplinary working	3.05
If informal carers have training in HD	3.00
Assessment of management of aggression if it occurs.	2.79
Having community understanding	2.74
Having a choice about seeing a male or a female GP	2.00

Cluster 9 for people with HD, Care Homes.

Two statements specifically relating to living in care homes shown in Table 4.2.9 comprised the ninth most important cluster for people with HD.

Table 4.2.9 Cluster 9, Care homes (Pw HD) Statements	Mean importance rating
Being able to go away from the care home to engage in community activities	2.68
Having meaningful relationships with other care home residents	1.89

Cluster 10 for people with HD, Coping by avoiding.

The grouping of statements concerned with smoking together with the suggestion that it is helpful not to know whether other family members are genetically positive for HD (Table 4.2.10) suggested to the steering group and validation group that people with HD regarded these as ways to cope by avoiding having to think about HD and its impact on the family. This was supported by notes from brainstorming sessions that recorded comments from some people with HD suggesting that smoking is an important factor in *'taking your mind off all these problems'.*

Table 4.2.10 Cluster 10, Coping by Avoiding (Pw HD) Statements	Mean importance rating
Not knowing whether other family members are genetically positive for HD	2.00
Having cigarettes	1.89
Having a smoking aid	1.63

Caregivers

The priority order of the 94 statements for the caregiver subgroup is shown in appendix 4.4. A ten-cluster solution was agreed and Fig 4.3 and Table 4.3 below demonstrate that the highest scoring cluster for caregivers was 'Long term planning'. Tables 4.3.1 to 4.3.10 present the statements making up the content of each caregiver cluster.

Fig 4.3 Concept map for caregivers

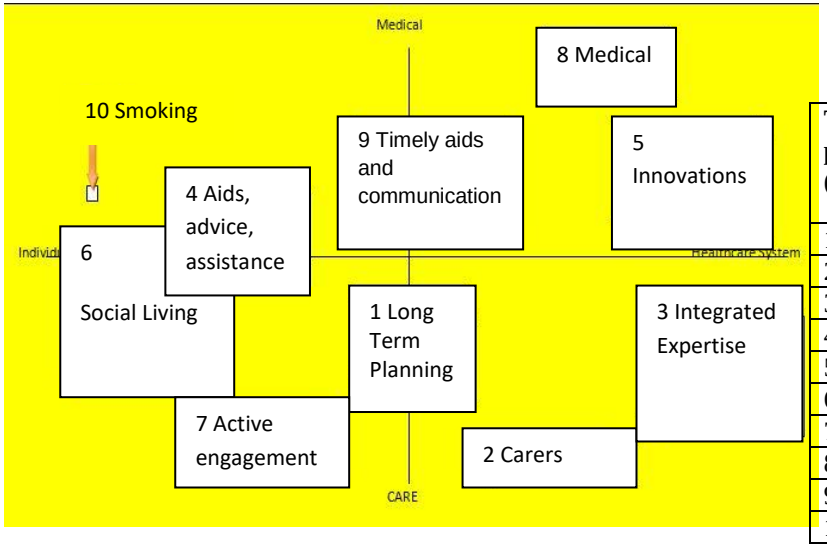


Table 4.3 Clusters for caregivers in priority order based on mean score (Cluster 1 = most important).	Mean importance rating
1 Long term planning	3.41
2 Carers	3.40
3 Integrated expertise	3.31
4 Accepting aids, advice and assistance	3.25
5 Access to innovations	2.99
6 Social living	2.93
7 Continuing active engagement	2.88
8 Medical input	2.76
9 Timely aids and communication	2.70
10 Smoking	1.46

The highest scoring cluster for caregivers, Long term planning' contains five statements referring to issues including support from the Huntington's Disease Association and choices about end of life care, as shown in Table 4.3.1 below. Tables 4.3.1 to 4.3.10 show the statements that make up each cluster in the cluster map for caregivers.

Cluster 1 for caregivers, Long term planning

The theme for the most important cluster for caregivers is long term planning. This links having support form the Huntington's Disease Association with statements relating to therapy to help with swallowing difficulties (a feature of advanced disease), regular assessment for aids and adaptations and training to use equipment, and choices about end of life care (Table 4.3.1).

Table 4.3.1 Cluster 1 for caregivers: Long term planning.	**Mean importance rating**
Having support from the Huntington's Disease Association.	3.88
Speech and language therapy involvement to help with swallowing problems	3.88
Regular timely assessment for aids and adaptations	3.38
Making choices about care including end of life care in advance and having the choice respected	3.16
Having appropriate training to use equipment	2.73

Cluster 2 for caregivers, 'Carers'.

This second most important cluster for family carers consists of three statements directly relating to the treatment of carers- being listened to and being able to take a break; to having help from visiting carers, paid and unpaid; and to suitability of a care home (Table 4.3.2). Incorporating the concern about quality of care homes into this cluster was considered by the validation group to infer that knowing a loved one is receiving good residential care helps carers to live with not having the person at home.

Table 4.3.2 Cluster 2 for caregivers 'Carers'.	**Mean importance rating**
If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.	4.19
If the family carer has a break from looking after him or her.	3.88
If carers are listened to as an adult and believed and respected	3.58
If a care home environment is made to feel as home should feel	3.15
If visiting carers are flexible regarding the specific type of care they provide	3.08
If professional carers are flexible about who they help in the household (for	2.54

example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).	

Cluster 3 for caregivers, Integrated expertise

Twenty three statements make up the third most important cluster for caregivers within which the steering and validation groups agreed the central theme is integrated expertise (Table 4.3.3).

Table 4.3.3 Cluster 3 for caregivers, Integrated expertise **Statements**	**Mean importance rating**
If care is available when the person with HD needs it.	4.23
Being referred to a specialist National HD centre	4.12
Access to professionals who are expert in HD	4.04
Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation	4.00
Having continuity of care staff, (e.g. professional carers in homes or that visit at home).	3.88
Access to a multi-disciplinary team that is expert in HD	3.77
Not being cared for in an inappropriate setting (e.g. A psychiatric or elderly setting where staff don't know about HD).	3.69
Having access to the right level of information about HD at the right time	3.62
If care is available where the person with HD needs it	3.62
If there is expert assessment and treatment of mental health problems (eg. depression, anxiety) occurring along with HD	3.58
If informal carers have training in HD	3.38
Continuity of health and social care professionals	3.35
Regular and timely access to health and social care assessment	3.27
If professionals help the person with HD to get a life	3.23
When service provision is planned for when it will be needed, rather than thought about once things have already got difficult	3.23
If professionals work with all members of the family	3.08
If there is information about HD for health professionals in general medical settings	3.08
Effective interagency working	2.77
If people with HD know the role of each professional	2.77
If professionals follow up patients who do not keep appointments	2.73
Effective multi-disciplinary working	2.65
Timeliness in access to care homes	2.50
Having a choice about seeing a male or a female GP	1.50

Cluster 4 for caregivers, Accepting aids, advice and assistance

The fourth most important cluster for caregivers contains five statements relating to the person with HD accepting aids, advice and assistance (Table 4.3.4). In

brainstorming sessions caregivers frequently mentioned that coping with HD is much easier if the person with the diagnosis accepts the need for help and advice and aids and equipment if these are needed.

Table 4.3.4 Cluster 4 for caregivers, Accepting aids, advice and assistance **Statements**	**Mean importance rating**
Having aids to enable daily living (e.g. Aids that help with communication, mobility, furniture, bathroom and falls prevention).	3.73
Having assistance to eat and drink	3.38
If they accept the diagnosis	3.27
If there is availability of food that is appetising and manageable	3.12
Practical advice on specific foods and textures	2.72

Cluster 5 for caregivers, Access to innovations

The theme of the fifth most important cluster for caregivers is having access to a broad range of innovations such as diagnosis, pre-implantation diagnosis, medication reviews, management of aggression, genetic counselling, and experimental treatments (Table 4.3.5).

Table 4.3.5 Cluster 5 for caregivers, Access to innovations	**Mean importance rating**
A prompt diagnosis	3.38
Having appropriately timed reviews of medication	3.20
Assessment of management of aggression if it occurs.	3.15
Having the opportunity to be involved in research	3.00
Pre-test and post-test counselling	3.00
Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).	2.65
Availability of pre-implantation diagnosis	2.58

Cluster 6 for caregivers, Social living

Cluster 6, sixth most important for caregivers (Table 4.3.6) is large in terms of the number of included statements (27) and is concerned with many aspects of social living.

Table 4.3.6 Cluster 6 for caregivers, Social living	Mean
Having a familiar daily routine.	4.19
Having a supportive network of family and friends	4.15
If the person with HD has help to be able to get out and about	4.04
Having a stable relationship with a partner	4.04
Having family and friends who offer practical support	4.04
If he or she is able to talk freely and openly about HD within the family	3.81
Having a positive outlook on life	3.62
Having help to socialise	3.46
Having opportunities for meaningful conversation	3.46
Participating in physical activities	3.31
Having someone to sort out his or her access to financial benefits	3.19
If they are listened to as an adult and believed and respected.	3.19
Living for today	3.00
Being able to take up hobbies and pastimes	2.92
Knowing in advance the detailed plans for what will happen today and / or at forthcoming events	2.77
Having accessible town, transport and shop facilities needed now	2.77
Having community understanding	2.69
Having practical strategies to enable him or her to eat out without embarrassment	2.54
Participating in intellectual activities	2.50
If they are enabled to maintain the roles that they feel suit their gender	2.35
If employers make reasonable adjustments to enable continued employment	2.31
Being able to undertake domestic activities such as cooking, gardening and cleaning	2.27
Spending money while he or she is able to	2.00
Having alcohol	1.88
Still being able to drive.	1.77
Having cigarettes	1.58
Being prudent with money	1.50

Cluster 7 for caregivers, Continuing active engagement.

The seventh most important cluster for caregivers is concerned with people with HD being able to actively engage within Huntington's Disease Association support groups, the community and within care homes (Table 4.3.7).

ITable 4.3.7 Cluster 7 for caregivers, Continuing active engagement	Mean
Being involved in a Huntington's Disease Association support group	3.85
Being able to go away from the care home to engage in community activities	2.65
Having meaningful relationships with other care home residents	2.15

Cluster 8 for caregivers, Medical input

Table 4.3.8 groups together 11 statements relating to various aspects of what the steering and validation groups saw as perceived by caregivers as medical input. This was rated the eighth most important cluster for caregivers.

Table 4.3.8 Cluster 8 for caregivers, Medical input.	**Mean**
Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living	3.50
Having medicines to help sleep	3.23
Medication against chorea	3.12
If obsessive behaviour and impulsivity are recognised as serious symptoms of HD	3.04
Having the goal of a normal life when considering the use of medication	3.00
The availability of a genetic test	2.92
Knowing that side effects of drugs for HD can be serious	2.54
Recognition that antipsychotic medication can be helpful in HD	2.42
Knowing that sedative medication will not be given solely to manage behaviour	2.31
Knowing that the effects of medications for HD are variable	2.27
Recognition that facial and neck pain can be a problem in HD.	2.00

Cluster 9 for caregivers, Timely aids and communication.

The ninth most important cluster for caregivers links five statements concerned with timely access to aids and with communication (Table 4.3.9).

Table 4.3.9 Cluster 9 for caregivers, Timely aids and communication **Statement**	**Mean importance rating**
Having timely access to appropriate aids and adaptations	3.58
Speech and language therapy involvement in maintaining ability to communicate	3.54
Timely introduction of tube feeding	2.46
Knowing if other family members are genetically positive for HD	2.00
Not knowing whether other family members are genetically positive for HD	1.92

Cluster 10 for caregivers, Smoking

The statement about having a smoking aid was isolated within its own cluster by caregivers, and rated as the least important cluster for this subgroup (Table 4.3.10).

Table 4.3.10 Cluster 10 for caregivers, Smoking **Statement**	**Mean importance rating**
Having a smoking aid	1.46

Health professionals

The priority order of the 94 statements for the health professional subgroup is shown in Appendix 4.5. A ten-cluster solution was agreed (see Fig 4.4 and Table 4.4). The highest scoring cluster for health professionals was 'Later stages' and Tables 4.4.1 to 4.4.10 below present the statements making up each cluster in priority order.

Fig 4.4 Cluster map for Health Professionals

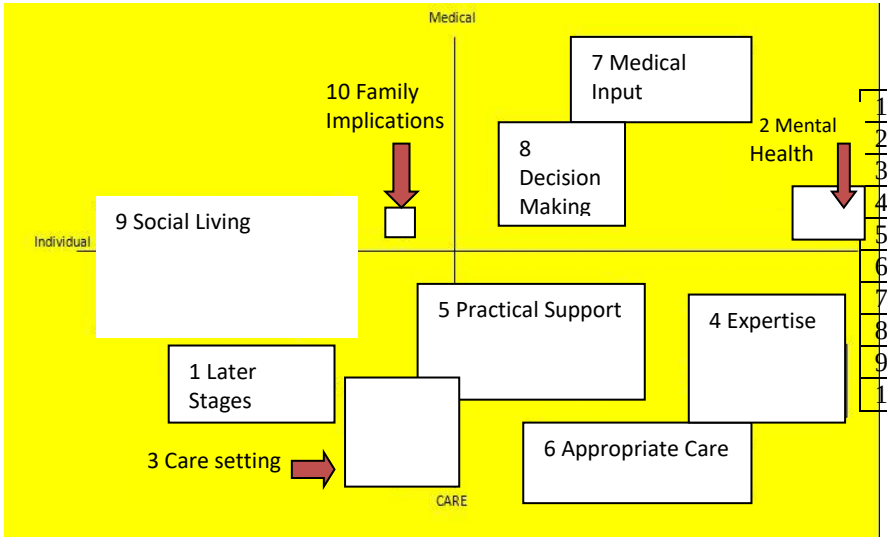


Table 4.4 Clusters for health professionals in priority order based on mean score (cluster 1 = most important).

1 Later stages	3.95
2 Co-morbid mental health problem	3.85
3 Care setting	3.33
4 Expertise	3.33
5 Practical support	3.31
6 Timely, flexible, appropriate care	3.20
7 Medical input	2.99
8 Decision-making	2.91
9 Social living	2.88
10 Family implications	1.92

The statements that make up each of the health professional clusters are shown in Tables 4.4.1 to 4.4.10.

Cluster 1 for health professionals, Later Stages.

The highest priority cluster for health professionals contains two statements that both relate to later stages of HD: end of life care and having assistance to eat and drink (Table 4.4.1).

Table 4.4.1 Cluster 1 for health professionals, Later Stages. **Statement**	**Mean Importance Rating**
Making choices about care including end of life care in advance and having the choice respected	4.15
Having assistance to eat and drink	3.75

Cluster 2 for health professionals, Mental health.

For health professionals the second highest rated cluster is made up of a single statement and is identical to the most highly rated cluster for combined subgroups, which prioritises having expert assessment and treatment of mental health problems occurring with HD (Table 4.4.2).

Table 4.4.2 Cluster 2 for health professionals, Mental Health. **Statement**	**Mean priority rating**
If there is expert assessment and treatment of mental health problems (e.g. Depression, anxiety) occurring along with HD	3.85

Cluster 3 for health professionals, Care setting

The cluster rated as third priority for health professionals contains three statements suggesting the importance of a care environment in which staff members have knowledge of and traing about HD and which is made to feel like home. Notes from brainstorming sessions recorded participants from all subgroups mentioning that

placement for people with HD in general psychiatric or elderly care settings was inappropriate (Table 4.4.3).

Table 4.4.3 Cluster 3 for health professionals, Care setting. **Statement**	**Mean priority rating**
Not being cared for in an inappropriate setting (eg. A psychiatric or elderly setting where staff don't know about HD).	3.90
If informal carers have training in HD	3.05
If a care home environment is made to feel as home should feel	3.05

Cluster 4 for health professionals, Expertise

The eleven statements making up the fourth priority cluster for health professionals highlights the importance of access to integrated team working among professionals with specialist HD expertise (Table 4.4.4).

Table 4.4.4 Cluster 4 for health professionals, Expertise.	**Mean priority rating**
Access to a multi-disciplinary team that is expert in HD	4.10
Access to professionals who are expert in HD	4.05
Regular and timely access to health and social care assessment	4.00
Effective multi-disciplinary working	3.70
Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation	3.65
Effective interagency working	3.30
Being referred to a specialist National HD centre	3.05
Continuity of health and social care professionals	3.05
If there is information about HD for health professionals in general medical settings	2.70
If professionals follow up patients who do not keep appointments	2.60
If people with HD know the role of each professional	2.45

Cluster 5 for health professionals, Practical support.

The steering and validation groups agreed that the theme of practical support is central to the twelve statements making up the fifth highest priority cluster for health professionals (Table 4.4.5).

Table 4.4.5 Cluster 5 for Health professionals, Practical support Item **Statement**	**Mean priority rating**
Having aids to enable daily living (e.g. aids that help with communication, mobility, furniture, bathroom and falls prevention).	4.00

Speech and language therapy involvement to help with swallowing problems	3.95
If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.	3.80
Having timely access to appropriate aids and adaptations	3.75
Regular timely assessment for aids and adaptations	3.60
Speech and language therapy involvement in maintaining ability to communicate	3.60
If the family carer has a break from looking after him or her.	3.60
If professionals help the person with HD to get a life	3.15
Having support from the Huntington's Disease Association.	3.10
Having access to the right level of information about HD at the right time	3.05
Having a choice about seeing a male or a female GP	2.15
Having appropriate training to use equipment	1.95

Cluster 6 for health professionals, Timely flexible, appropriate care.

The importance of timeliness and flexibility and the appropriateness of care for people with HD is expressed in the sixth highest rated cluster for health professionals, containing eight statements (Table 4.4.6).

Table 4.4.6 Cluster 6 for health professionals, Timely, flexible, appropriate care	**Mean**
If care is available when the person with HD needs it	3.85
When service provision is planned for when it will be needed, rather than thought about once things have already got difficult	3.80
If care is available where the person with HD needs it.	3.65
If professionals work with all members of the family	3.20
Having continuity of care staff, (e.g., professional carers in homes or that visit at home)	3.00
Timeliness in access to care homes	2.80
If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).	2.70
If visiting carers are flexible regarding the specific type of care they provide	2.60

Cluster 7 for health professionals, Medical input.

The theme of medical input was not given as high a priority rating by health professionals (seventh highest rated cluster) as by people with HD for whom it was the second highest aspect of what helps to live with the condition.

Table 4.4.7 Cluster 7 for health professionals, Medical input **Statement**	**Mean priority rating**
Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.	4.15

Medication against chorea	3.90
Having appropriately timed reviews of medication	3.65
Having medicines to help sleep	3.50
Having the goal of a normal life when considering the use of medication	3.25
Recognition that antipsychotic medication can be helpful in HD	3.20
Assessment of management of aggression if it occurs.	3.20
Knowing that sedative medication will not be given solely to manage behaviour	2.70
If obsessive behaviour and impulsivity are recognised as serious symptoms of HD	2.70
Knowing that side effects of drugs for HD can be serious	2.35
Knowing that the effects of medications for HD are variable	2.30
Recognition that facial and neck pain can be a problem in HD.	2.05
Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).	1.90

Cluster 8 for health professionals, Decision-making.

This eighth most important cluster from the health professional viewpoint brings together issues such as diagnosis, genetic testing and related counselling, whether to take part in research, and whether to introduce tube feeding in response to swallowing problems, that can often present people with HD and their families and also health professionals with diffcult choices that must be made (Table 4.4.8).

Table 4.4.8 Cluster 8 for health professionals, Decision-making. **Statement**	**Mean**
Pre-test and post-test counselling	3.50
A prompt diagnosis	3.35
Timely introduction of tube feeding	2.95
Availability of pre-implantation diagnosis	2.75
The availability of a genetic test	2.65
Having the opportunity to be involved in research	2.2

Cluster 9 for health professionals, Social living.

Health professionals formed one very large (36 statements) cluster which they rated ninth most important, the themes of which was agreed by the steering and validation groups to be social living (Table 4.4.9).

Table 4.4. 9 Cluster 9 for health professionals, Social living **Statement**	**Mean importance rating**
If they are listened to as an adult and believed and respected.	4.35
Having a familiar daily routine.	4.10
Having a supportive network of family and friends	4.00
If he or she is able to talk freely and openly about HD within the family	3.90
Having opportunities for meaningful conversation	3.75
Having family and friends who offer practical support	3.70
Knowing in advance the detailed plans for what will happen today and / or at forthcoming events	3.40
Having a stable relationship with a partner	3.35
If employers make reasonable adjustments to enable continued employment	3.30
If the person with HD has help to be able to get out and about	3.25
If carers are listened to as an adult and believed and respected	3.25
Living for today	3.15
Being involved in a Huntington's Disease Association support group	3.15
If there is availability of food that is appetising and manageable	3.10
If they accept the diagnosis.	3.05
Having practical strategies to enable him or her to eat out without embarrassment	3.05
Being able to go away from the care home to engage in community activities	3.05
Having a positive outlook on life	3.00
Having help to socialise	2.95
Having community understanding	2.90
Practical advice on specific foods and textures	2.90
If they are enabled to maintain the roles that they feel suit their gender	2.85
Having someone to sort out his or her access to financial benefits	2.65
Having accessible town, transport and shop facilities	2.60
Being able to undertake domestic activities such as cooking, gardening and cleaning	2.60
Direct access to cash benefits to spend on what is needed now	2.55
Participating in physical activities	2.50
Participating in intellectual activities	2.35
Being able to take up hobbies and pastimes	2.30
Having cigarettes	2.10
Having a smoking aid	1.95
Having meaningful relationships with other care home residents	1.90
Spending money while he or she is able to	1.85
Still being able to drive.	1.85
Being prudent with money	1.65
Having alcohol	1.50

Cluster 10, Family implications.

Two polarised statements concerned with whether knowing or not knowing if other family members are genetically positive for HD is helpful were grouped together by health professionals in the cluster they rated as least important (Table 4.4.10).

Table 4.4.10 Cluster 10 for health professionals, Family implications. **Statement**	**Mean importance rating**
Knowing if other family members are genetically positive for HD	2.20
Not knowing whether other family members are genetically positive for HD	1.65

Nursing home-based subgroup

The nursing home-based subgroup consisted of 19 Caregivers and eight health professionals who identified themselves as having their main involvement with HD relating to a person with the condition who resides at a nursing home or similar long term care facility. Each of the people with HD who resided at a nursing home and who participated in brainstorming was unable to underetake the more onerous tasks of prioritising and clustering statements. The priority order of the 94 statements for the 'nursing home-based subgroup' is shown in Appendix 4.6. An eight-cluster solution was agreed and Table 4.5 below demonstrates that the highest scoring cluster for the nursing home-based subgroup was 'Care'.

Fig 4.5 Cluster map for the nursing home-based subgroup

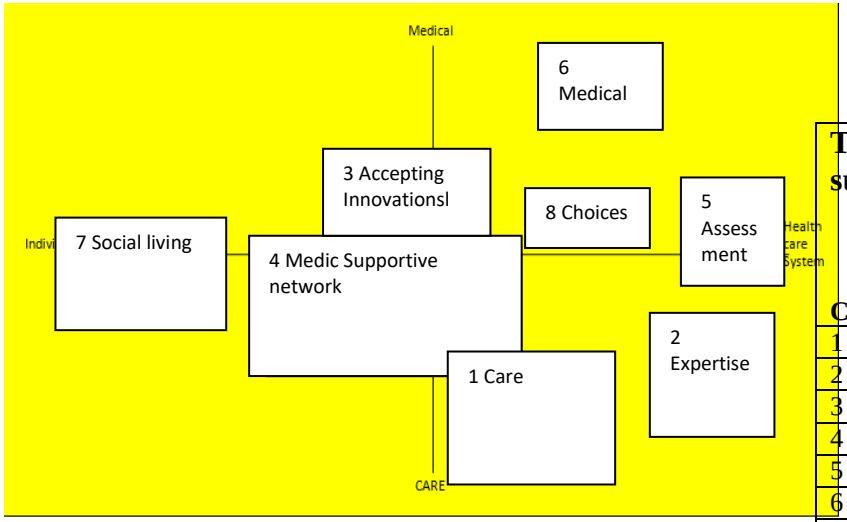


Table 4.5 Clusters for the Nuring home-based subgroup, Care **Cluster Name**	**Mean importance rating (cluster 1 = most important)**
1 Care	3.30
2 Expertise	3.29
3 Accepting innovations	3.27
4 Interaction with a supportive network	3.21
5 Assessment by informed professionals	3.12
6 Medical input	3.00
7 Social living	2.75
8 Choices about diagnosis & consulting	2.68

The statements that make up each of the nursing home-based clusters are shown in Tables 4.5.1 to 4.5.8.

Cluster 1 for the nursing home-based subgroup

Fourteen statements make up the highest scoring (most important) cluster ('care') for the nursing home-based subgroup (Table 4.5.1).

Table 4.5.1 Cluster 1 for the nursing home-based subgroup, Care.	**Mean importance rating**
If care is available where the person with HD needs it.	3084
If care is available when the person with HD needs it.	3.84
If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.	3.84
Not being cared for in an inappropriate setting (eg. A psychiatric or elderly setting where staff don't know about HD).	3.63
Being involved in a Huntington's Disease Association support group	3.58
If the family carer has a break from looking after him or her.	3.58
Having continuity of care staff, (e.g. professional carers in homes or that visit at home).	3.42
Having support from the Huntington's Disease Association.	3.32
If professionals help the person with HD to get a life	3.26
If a care home environment is made to feel as home should feel	3.16
If informal carers have training in HD	2.84
If visiting carers are flexible regarding the specific type of care they provide	2.84
If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).	2.58
Timeliness in access to care homes	2.47

Cluster 2 for the Nursing Home-based subgroup, Care.

Expertise is the central theme of the twelve-statement cluster rated as second most important for the nursing home-based subgroup (Table 4.5.2). The aspects rated as most important within the cluster are having access to a multidisciplinary team and to individual professionals with HD expertise, and referral to a national HD centre.

Table 4.5.2 Cluster 2 for the nursing home-base subgroup, Expertise.	Mean
Access to a multi-disciplinary team that is expert in HD	4.32
Access to professionals who are expert in HD	4.21
Being referred to a specialist National HD centre	3.74
When service provision is planned for when it will be needed, rather than thought about once things have already got difficult	3.58
Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation	3.53
Having access to the right level of information about HD at the right time	3.42
Effective multi-disciplinary working	3.00
Continuity of health and social care professionals	3.00
Effective interagency working	2.84
If professionals work with all members of the family	2.84
If professionals follow up patients who do not keep appointments	2.63
If people with HD know the role of each professional	2.42

Cluster 3 for the nursing home-based subgroups, Accepting innovations.

The steering and validation groups found the theme of this cluster, that is the factor that caused members of this subgroup to frequently place these six statements together to be less obvious than was the case for most clusters, but they agreed that acceptance by people with HD of a variety of innovations could be seen as a common thread (Table 4.5.3).

Table 4.5.3 Cluster 3 for the nursing home-based subgroup, Accepting innovations.	Mean
If they accept the diagnosis.	3.68
Speech and language therapy involvement in maintaining ability to communicate	3.58
Having timely access to appropriate aids and adaptations	3.42
Regular timely assessment for aids and adaptations	3.37
Timely introduction of tube feeding	3.16
Having the opportunity to be involved in research	2.42

Cluster 4 for the nursing home-based subgroup, Interaction with a supportive network.

The grouping of the fourteen statements in cluster 4 was more easily understood by the steering and validation groups (see comments, cluster 3, above), and the theme of interaction with a supportive network was identified (Table 4.5.4).

Table 4.5.4 Cluster 4 for the nursing home-based subgroup, Interaction with a supportive network.	**Mean importance rating**
Having a supportive network of family and friends	4.05
Speech and language therapy involvement to help with swallowing problems	3.95
If he or she is able to talk freely and openly about HD within the family	3.89
Having aids to enable daily living (eg. aids that help with communication, mobility, furniture, bathroom and falls prevention).	3.89
Having assistance to eat and drink	3.79
If they are listened to as an adult and believed and respected.	3.63
Having family and friends who offer practical support	3.58
If carers are listened to as an adult and believed and respected	3.53
Making choices about care including end of life care in advance and having the choice respected	3.11
Practical advice on specific foods and textures	3.00
Knowing if other family members are genetically positive for HD	2.32
Having appropriate training to use equipment	2.11
Having meaningful relationships with other care home residents	2.11
Not knowing whether other family members are genetically positive for HD	1.95

Cluster 5 for the nursing home-based subgroup, Assessment by informed professionals.

Two statements concerned with the value of assessment which should be regular and timely, and by experts; and a third promoting the importance of professionals being informed about HD make up this cluster ranked as fifth priority for the nursing home-based subgroup (Table 4.5.5).

Table 4.5.5 Cluster 5 for the nursing home-based subgroup, Assessment by informed professionals. **Statement**	**Mean importance rating**
Regular and timely access to health and social care assessment	3.42
If there is expert assessment and treatment of mental health problems (e.g. depression, anxiety) occurring along with HD	3.32
If there is information about HD for health professionals in general medical settings	2.63

Cluster 6 for the nursing home-based subgroup, Medical input.

Thirteen statements with various aspects of medical input were combined in the sixth most important cluster for the nursing home-based subgroup (Table 4.5.6).

Table 4.5.6 Cluster 6, for the nursing home-based subgroup, Medical input.	**Mean importance rating.**
Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.	4.11
Medication against chorea	3.89
Having medicines to help sleep	3.42
Recognition that antipsychotic medication can be helpful in HD	3.42
Having the goal of a normal life when considering the use of medication	3.32
Having appropriately timed reviews of medication	3.26
Assessment of management of aggression if it occurs.	3.16
If obsessive behaviour and impulsivity are recognised as serious symptoms of HD	2.74
Knowing that sedative medication will not be given solely to manage behaviour	2.63
Knowing that the effects of medications for HD are variable	2.32
Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).	2.32
Recognition that facial and neck pain can be a problem in HD.	2.26
Knowing that side effects of drugs for HD can be serious	2.21

Cluster 7 for the nursing home-based subgroup, Social living.

The seventh most important aspect of help for people living with HD from the nursing home-based perspective is captured in a large cluster of 27 statements with social living as a central theme (Table 4.5.7).

Table 4.5.7 Cluster 7 for the nursing home-based subgroup, Social living **Statement**	**Mean importance rating**
Having a familiar daily routine.	4.26
Having a stable relationship with a partner	3.68
If the person with HD has help to be able to get out and about	3.63
Having opportunities for meaningful conversation	3.37
Having help to socialise	3.21
Knowing in advance the detailed plans for what will happen today and / or at forthcoming events	3.21
If there is availability of food that is appetising and manageable	3.16
Participating in physical activities	3.16
If employers make reasonable adjustments to enable continued employment	3.05
Being able to go away from the care home to engage in community activities	3.05
Having a positive outlook on life	2.94
Living for today	2.89
Having accessible town, transport and shop facilities	2.84
Having practical strategies to enable him or her to eat out without embarrassment	2.74
Having someone to sort out his or her access to financial benefits	2.68
Having community understanding	2.63
If they are enabled to maintain the roles that they feel suit their gender	2.63
Being able to take up hobbies and pastimes	2.63
Direct access to cash benefits to spend on what is needed now	2.53
Being able to undertake domestic activities such as cooking, gardening and cleaning	2.37
Participating in intellectual activities	2.32

Having cigarettes	2.11
Still being able to drive.	2.11
Spending money while he or she is able to	1.84
Having a smoking aid	1.84
Being prudent with money	1.79
Having alcohol	1.68

Cluster 8 for the nursing home-based subgroup, Choices about diagnosis and consulting.

The theme within the cluster rated eighth and least important by the nursing home-based subgroup consists of five statements that combine factors relating to making choices about diagnosis and counselling.

Table 4.5.8 Cluster 8 for the nursing home-based subgroup, Choices about diagnosis & consulting **Statement**	**Mean**
A prompt diagnosis	3.05
Pre-test and post-test counselling	3.00
Availability of pre-implantation diagnosis	2.89
The availability of a genetic test	2.26
Having a choice about seeing a male or a female GP	2.21

Community-based subgroup

Seven caregivers and 12 health professionals identified their involvement with HD as community-based as opposed to nursing-home based. It is acknowledged that the distinction is imperfect as within one family there may be close members living at home and in a nursing home. The priority order of the 94 statements for the 'community-based subgroup is shown in Appendix 4.6. A ten-cluster solution was agreed and Fig 4.6 and Table 4.6 below demonstrate that the highest scoring cluster for the community-based subgroup was 'avoiding crises'.

Fig 4.6 Concept map for the community-based subgroup

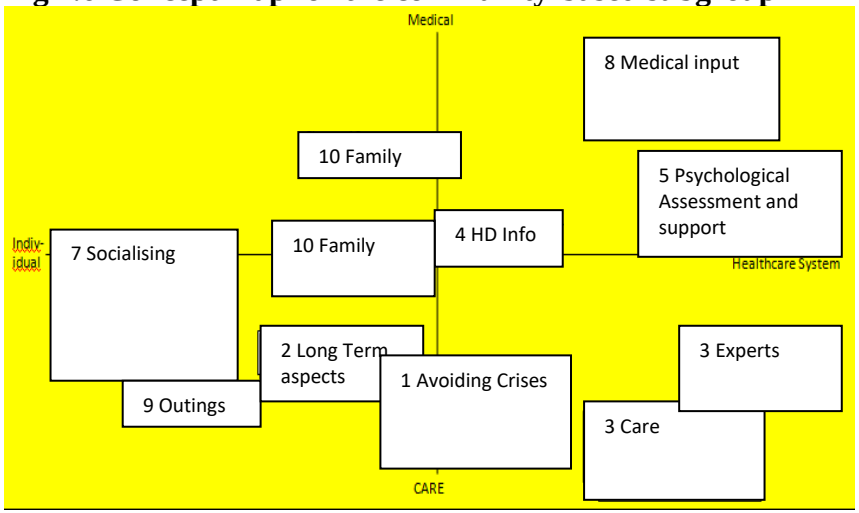


Table 4.6 Clusters for the community-based subgroup in priority order based on mean score (cluster 1 = most important). (N=26; caregivers-17; HPs-9) **Cluster Name**	**Mean importance rating**
1 Avoiding Crises	3.59
2 Longer Term Aspects	3.46
3 Care	3.40
4 Information About HD	3.36
5 Psychological Assessment and Support	3.27
6 Expert Professionals	3.20
7 Social living	2.86
8 Medical input	2.82
9 Outings	2.80
10 Family implications	1.86

The statements that make up each of the community-based clusters are shown in tables 4.6.1 to 4.6.10

Cluster 1 for the community-based subgroup, Avoiding crises.

The highest scoring (most important) cluster for the community subgroup contains nine statements concerned with avoidance of problems such as carers reaching a tipping point beyond which they cannot cope and swallowing problems in people with HD; and with accessing assessment and aids and adaptations at a time when they are needed. The theme was considered by the steering and validation groups to be 'avoiding crises' (Table 4.6.1).

Table 4.6.1 Cluster 1 for the community-based subgroup, Avoiding crises	**Mean importance rating**
If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.	4.15
Speech and language therapy involvement to help with swallowing problems	3.96
If the family carer has a break from looking after him or her.	3.88
Having timely access to appropriate aids and adaptations	3.85
Having aids to enable daily living (eg. aids that help with communication, mobility, furniture, bathroom and falls prevention).	3.77
Regular timely assessment for aids and adaptations	3.58
Speech and language therapy involvement in maintaining ability to communicate	3.54
If a care home environment is made to feel as home should feel	3.12
Having appropriate training to use equipment	2.50

Cluster 2 for the community-based subgroup, Longer term aspects.

The second most important cluster for the community-based subgroup draws together factors determined by the steering and validation groups as 'longer term aspects' such as end of life care choices, support from the Huntington's Disease Association, carers being believed and listened to and having assistance to eat and drink (Table 4.6.2).

Table 4.6.2 Cluster 2 for the community-based subgroup, Longer term aspects. Statements	Mean Importance rating
Making choices about care including end of life care in advance and having the choice respected	4.00
Having support from the Huntington's Disease Association.	3.69
If carers are listened to as an adult and believed and respected	3.42
Having assistance to eat and drink	3.31
Practical advice on specific foods and textures	2.72

Cluster 3 for the Community-based subgroup, Care.

Cluster 3 for the community-based subgroup combines 12 statements relating to assessment for, access to and provision of care (Table 4.6.3).

Table 4.6.3 Cluster 3 for the Community-based subgroup, Care. Statement	Mean priority rating
If care is available when the person with HD needs it.	4023
Not being cared for in an inappropriate setting (e.g. A psychiatric or elderly setting where staff don't know about HD).	3.96
Regular and timely access to health and social care assessment	3.77
Having continuity of care staff, (e.g. professional carers in homes or that visit at home).	3.58
If informal carers have training in HD	3.54
When service provision is planned for when it will be needed, rather than thought about once things have already got difficult	3.46
If care is available where the person with HD needs it.	3.46
If professionals work with all members of the family	3.42
Effective multi-disciplinary working	3.27
If visiting carers are flexible regarding the specific type of care they provide	2.85
Timeliness in access to care homes	2.81
If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).	2.62

Cluster 4 for the Community-based subgroup, Information about HD.

A single statement about having access to an appropriate level of information about HD constitutes the fourth priority cluster for the community-based subgroup (Table 4.6.4).

Table 4.6.4 Cluster 4 for the Community-based subgroup, Information about HD StatementItem	Mean priority rating
Having access to the right level of information about HD at the right time	3.38

Cluster 5 for the community-based subgroup, Psychological assessment and support.

The fifth priority cluster for the community-based subgroup contains 4 statements that refer to expert assessment of mental health problems, pre-and post genetic test counselling, management of aggression if this occurs and to timeliness of the introduction of tube feeding as helpful factors in living with HD (Table 4.6.5). The cluster was named 'Psychological assessment and support.

Table 4.6.5 Cluster 5 for the community-based subgroup, Psychological assessment and support. Statement	Mean priority rating
If there is expert assessment and treatment of mental health problems (e.g. Depression, anxiety) occurring along with HD	3.96
Pre-test and post-test counselling	3.38
Assessment of management of aggression if it occurs.	3.19
Timely introduction of tube feeding	2.38

Cluster 6 for the community-based subgroup, Expert professionals.

Eleven statements combine various aspects of expertise and also some of the attributes considered particularly helpful among professionals and underline the importance of access to this (Table 4.6.6). This cluster was rated as sixth most important for the community-based subgroup.

Table 4.6.6 Cluster 6 for the Community-based subgroup, Expert professionals Statement	Mean importance rating
Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation	4.04
Access to professionals who are expert in HD	3.88

Access to a multi-disciplinary team that is expert in HD	3.73
Being referred to a specialist National HD centre	3.62
Continuity of health and social care professionals	3.38
Effective interagency working	3.19
If professionals help the person with HD to get a life	3.12
If there is information about HD for health professionals in general medical settings	3.08
If people with HD know the role of each professional	2.85
If professionals follow up patients who do not keep appointments	2.77
Having a choice about seeing a male or a female GP	1.46

Cluster 7 for the Community-based subgroup, Social living

This large cluster for the community-based subgroup was rated as seventh most important, containing 32 statements all seen by the steering and validation groups to relate to some aspect of social living (Table 4.6.7).

Table 4.6.7 Cluster 7 for the Community-based subgroup, Social living **Statement**	**Mean priority rating**
Having a familiar daily routine.	4.12
Having family and friends who offer practical support	4.12
Having a supportive network of family and friends	4.08
Having a stable relationship with a partner	3.88
If he or she is able to talk freely and openly about HD within the family	3.88
If they are listened to as an adult and believed and respected.	3.81
If the person with HD has help to be able to get out and about	3.69
Having opportunities for meaningful conversation	3.69
Having a positive outlook on life	3.58
Being involved in a Huntington's Disease Association support group	3.50
Having help to socialise	3.19
Living for today	3.12
Having someone to sort out his or her access to financial benefits	3.12
If there is availability of food that is appetising and manageable	3.12
If they accept the diagnosis.	2.88
Having community understanding	2.81
Having practical strategies to enable him or her to eat out without embarrassment	2.77
Participating in physical activities	2.73
Direct access to cash benefits to spend on what is needed now	2.65
Having accessible town, transport and shop facilities	2.58
If they are enabled to maintain the roles that they feel suit their gender	2.58
If employers make reasonable adjustments to enable continued employment	2.58
Being able to take up hobbies and pastimes	2.58
Being able to undertake domestic activities such as cooking, gardening and cleaning	2.50
Participating in intellectual activities	2.42
Having meaningful relationships with other care home residents	2.00
Spending money while he or she is able to	1.92
Having alcohol	1.73
Having cigarettes	1.62
Still being able to drive.	1.62
Having a smoking aid	1.58
Being prudent with money	1.38

Cluster 8 for the community-based subgroup, Medical input

The central theme for the eighth most important cluster for the community-based subgroup is 'Medical input' and is made up of 16 statements (Table 4.6.8).

Table 4.6.8 Cluster 8 for the Community-based subgroup, Medical input **Statement**	**Mean priority rating**
A prompt diagnosis	3.62
Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.	3.50
Having appropriately timed reviews of medication	3.48
Having medicines to help sleep	3.23
The availability of a genetic test	3.19
Medication against chorea	3.08
If obsessive behaviour and impulsivity are recognised as serious symptoms of HD	3.00
Having the goal of a normal life when considering the use of medication	3.00
Having the opportunity to be involved in research	2.77
Knowing that side effects of drugs for HD can be serious	2.65
Availability of pre-implantation diagnosis	2.42
Knowing that sedative medication will not be given solely to manage behaviour	2.38
Recognition that antipsychotic medication can be helpful in HD	2.31
Knowing that the effects of medications for HD are variable	2.27
Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).	2.23
Recognition that facial and neck pain can be a problem in HD.	1.80

Cluster 9 for the community-based subgroup, Outings

Table 4.61 shows two statements concerned with going out within one cluster rated by the community-based subgroup as ninth in importance. Knowing plans in advance and being able to go out from a nursing home to engage in the community are highlighted helpful factors towards living with HD.

Table 4.6.9 Cluster 9 for the Community-based subgroup, Outings **Statement**	**Mean importance rating**
Knowing in advance the detailed plans for what will happen today and / or at forthcoming events	2.96
Being able to go away from the care home to engage in community activities	2.62

Cluster 10 for the Community-based subgroup, Family implications.

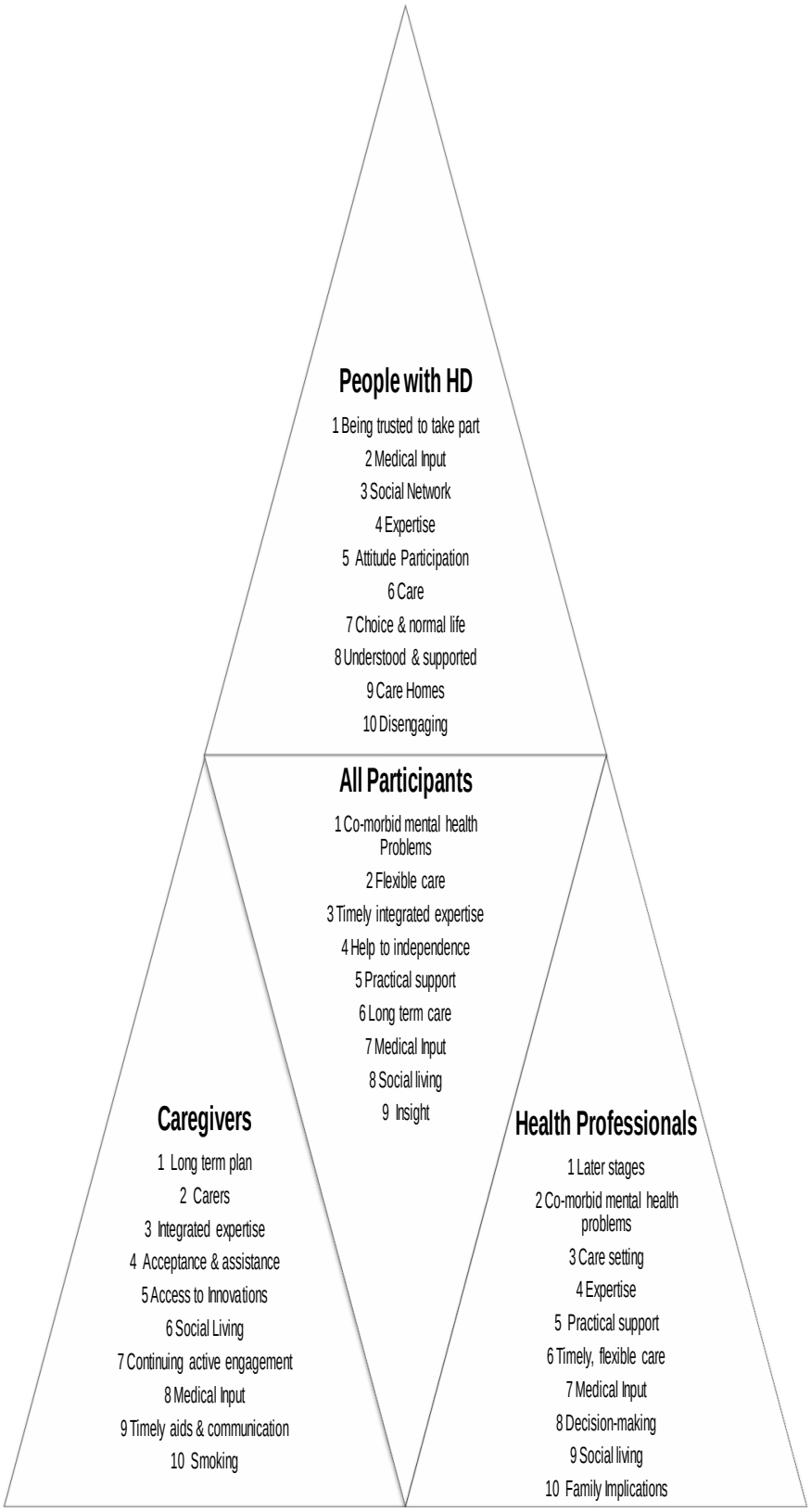
The cluster rated as least important for the community-based subgroup contains two statements indicating that both knowing and not knowing if other family members are genetically positive for HD can be helpful towards living with HD (Table 4.6.10).

Table 4.6.10 Cluster 10 for the Community-based subgroup, Family implications **Statement**	**Mean priority rating**
Knowing if other family members are genetically positive for HD	1.96
Not knowing whether other family members are genetically positive for HD	1.73

Comparing Subgroups

To further assist comparison of the clusters formed and prioritised by people with HD, caregivers and health professionals Fig 4.7 presents the clustering by all participants and by each of these three subgroups within one diagram, highest prioritised cluster for each subgroup is uppermost and numbered ‘1’.

Fig 4.7: Diagrammatic representation of cluster names for 'all participants' and for three subgroups



Contrasting values identified by the primary investigator, and ratified by the steering group and validation group were considered to capture similarities and differences between three subgroups: people with HD, caregivers and health professionals (see Table 4.7).

Table 4.7 Contrasting values between subgroups

Carers / Professionals Forward planning		**PWHD** Living for today
PWHD / Professionals Trusting the patient Confidentiality between Patient and Professional		**Carers** Recognising impaired insight Professionals liaising with carer
Carers Patient acceptance of condition Patient provided for Patient submission to care	**Professionals** The professional perspective regarding these aspects seems to be positioned between carer and people with HD perspectives	**PWHD** Patient autonomy Normal living Making choices Being understood Supported, not controlled
PWHD Medical solution emphasis		**Carers / Professionals** Care solution emphasis

Latent preferences

Figs 4.8-4.10 show latent preferences for people with HD, caregiver and health professional groups respectively to allow comparison (see methods, 3.10.8).

Fig 4.8 Concept map showing latent preferences for people with HD

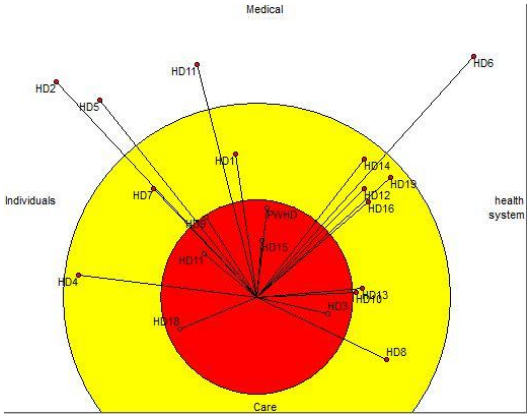
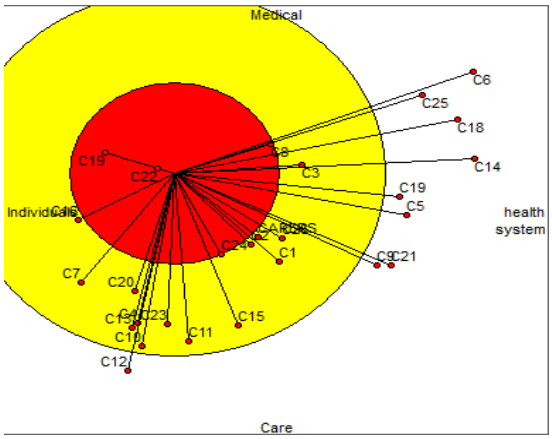


Fig 4.8 demonstrates a tendency for people with HD to prioritise statements relating to medical input, the health system and the individual and not to those relating to care.

Fig 4.9 Concept map showing latent preferences for caregivers



In contrast to Fig 4.8, Fig 4.9 demonstrates a greater tendency among caregivers to prioritise statements relating to care and the health system rather than to medical input or the individual.

Fig 4.10 Concept map showing latent preferences for health professionals.

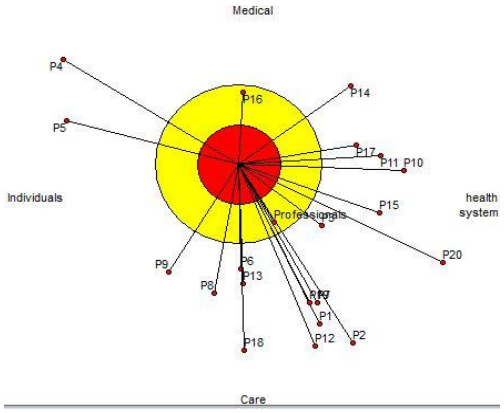


Fig 4.10 shows that with the exception of two outliers (coded P4 and P5) prioritising among health care professionals places greater emphasis on care than is placed by people with HD and greater emphasis on medical input than caregivers. The only professionals tending to prioritise statements concerned with individuals are the only two social workers who both also own and manage care homes.

Significant Differences

Results from the Kruskall Wallis H non-parametric ANOVA test showed that of the 94 items three were associated with a significant difference in how they were rated by subgroups as presented below in tables 4.8.

Table 4.8 Statements associated with significant difference in rating by subgroups at p=0.01

Statement	Most valued by	Least valued by	Subgroup mean importance rating	Kruskall-Wallis result for significance at p<0.01.
Having timely access to appropriate aids and adaptations	Professionals	People with HD	Profs: 3.75, Carers: 3.58 PwHD: 2.37	P=0.002
Still being able to drive	People with HD	Caregivers	PwHD: 3.42 Profs: 1.85 Carers: 1.77	P=0.001
Access to experimental treatments (not as part of a research study)	People with HD	Professionals	PwHD: 3.16 Carers: 2.65 Profs: 1.90	P=0.008

Nursing Home-Based and Community-Based Subgroup Comparison

Of the 94 statements, statistical difference ($p<0.01$) in ranking between the community-based and the nursing home-based subgroups occurred in one case, that of the statement '*What helps people with Huntington's disease live with their condition is recognition that antipsychotic medication can be helpful in HD.*' This statement was given a higher (more important) rating by nursing home-based caregivers and health professional participants (3.42) than by caregivers and health professionals in the community-based subgroup (2.31).

Caution should be exercised in interpreting this difference given that there are close to 100 statements making the 1% probability of occurrence in one case likely purely by chance. Table 4.16 shows the table produced by SPSS analysis using the Mann Whitney U test comparing distribution between independent samples. Two other statements were highlighted as associated with significant difference at $p<0.05$, but this higher probability of these results occurring by chance means they cannot be accepted as significant.

Having a positive outlook, and availability of genetic testing were given a higher mean rating by community-based (3.58 and 3.19 respectively) compared to nursing home-based participants (2.94 and 2.26 respectively) but the difference was not significant at $p=<0.01$ (Table 4.9).

Table 4.9 Differences between nursing home-based and community-based ratings of three statements

Statement *'What helps people with HD live with their condition is…*	**Level of significance** **(difference between nursing home-based and community-based subgroup responses)**	**Context** **(where rated as more important)**
…recognition that antipsychotic medication can be helpful in HD'.	P=0.008*	Nursing home
…having a positive outlook on life'.	P=0.049	Community
…availability of a genetic test'.	P=0.047	Community

Three statements in Table 4.16 were identified as 'significant' using Mann Whitney at $p<0.05$. For reasons explained above only significance at $p=<0.01$ is considered acceptable and therefore only the statement concerning recognition that antipsychotic medication can be helpful in HD, at ($p=0.008$) is viewed as significant.

Chapter Five
Discussion

5.1 Introduction

This is the first study providing a concept map based on quantitative analysis of qualitative statements generated by people with HD, family caregivers and health professionals with HD experience, in answer to the question 'what helps people with HD live with their condition?' The labels of the concept map axis are in keeping with findings of others who have used concept mapping to elicit, conceptualise and quantitatively rate subjective opinions of people with long term conditions, carers and professionals. Flaherty (2014) studied coeliac disease, and found that the process of plotting statements on the matrix tended to polarise issues to do with medical input versus aspects of care or management, and personal, individual factors from those to do with accessing the healthcare system.

5.2 Discussion of findings

The principle result is a combined opinion that *'expert assessment and treatment for co-morbid mental health problems occurring with HD'* is what helps most in living with HD. A criticism of this finding might be that the rating for the isolated statement constitutes the mean importance rating for the cluster, whilst other clusters contain statements with higher ratings. The fact that the statement became isolated as a cluster of its own suggests participants see it as an issue separate from the other themes that emerged. Ratification by the validation group, and feedback received from HD support group members indicating, that the cluster does capture the aspect of HD most difficult to manage, and for which alleviation would help most towards living with the condition, adds to my confidence in the validity of this cluster's prioritisation. Furthermore this finding resonates with research in long term

conditions which consistently highlights the high impact of mental health issues. Mental health problems occurring with long term physical symptoms are associated with significantly poorer health outcomes and a lower quality of life (Naylor et al 2012). Neurological disorders generally have a high prevalence of a variety of mental health problems and there is a close relationship between mental and physical wellbeing Royal College of Psychiatrists (2009). There is an increased level of disability in neurological conditions when compounded by psychiatric symptoms and these aspects add greatly to stress experienced by caregivers and are a significant factor contributing to reasons for resorting to institutional care World Health Organisation (2006).

The findings add weight to the suggestion Gudesblatt and Tarsy (2011) that an emphasis in HD research on motor symptoms may be misplaced and that a more integrated mental health support approach is needed (Naylor et al 2012). The Royal College of Psychiatrists (2009) recommends that all national guidelines for long term physical conditions should include specific advice about the detection and treatment of co-morbid mental health problems, and The Kings Fund proposes that care for people with co-morbid mental and physical health conditions should be a top ten priority for service providers (Imison et al 2011).

Clusters 2, 3 and 4 highlight the importance of flexibility in the delivery of appropriate and expert care. Hence access to HD specialist multidisciplinary care teams is of high importance to the participants. These findings are in keeping with guidelines and research in long term conditions (Department of Health 2005; Aspinal et al 2011) which highlight the importance of person-centred services, involving holistic integrated and expert multi-disciplinary and multi-agency care planning, review and service delivery in order to meet individual and changing

needs. Unfortunately this is not how current care is organised and implementing changes to meet these aspirations has proved extremely difficult (Sixsmith et al, 2013). Changes to professional attitudes and ways of working can be as important as changes to services (Satink et al 2014; Thomas et al 2010). Prompt referral to specialists with neurological expertise and integrated assessment and care planning are considered essential within the National Service Framework (Department of Health, 2005). The participants in *this* study went further, calling for neurological expertise to be HD specific. The willingness to travel long distances to access such expertise, and the positive change to managing life with HD experienced by those who have managed to do so is noteworthy. Continued support for concentrated centres of excellence is essential but they must also establish ways to devolve their expertise to localised interdisciplinary teams.

In delivering care, services and treatment it is necessary to be aware of differences in perspectives of people with HD, caregivers and health professionals. It is important to seek the opinions of people with HD with the caveat that the nature of the condition may impair insight, making the perspective of caregivers equally valuable. Professionals in this study placed a high value on listening to and believing patients, whereas carers expressed frustration at difficulty in having their view considered as equally relevant. In the context of decision-making concerning artificial hydration for dementia patients, nurse-physician communication has been identified as the most important factor for optimal care (Bryon et al 2008). In domestic settings family members assume the care role and where patient insight may be compromised it is difficult to see why carer-professional communication shouldn't be held with the same high regard when difficult care or treatment decisions must be reached. However, professionals should also keep in mind that some family caregivers may

inherit the condition and that subtle cognitive changes can occur many years prior to diagnosis and impair insight and judgement (Paulsen et al 2007). Unless impaired capacity is established in the person with HD, choice is a right (Katona and Livingston 2013), and even if capacity is impaired, it may be counter-productive to highlight the fact. However the insight and expertise of carers should also be respected and integrated into care plans, otherwise professionals risk overloading carers' capacity to cope with their role.

Indeed, the problem of impaired insight is a central theme connecting the four statements associated with significant differences of opinion between subgroups. Professionals need to consider that impairment exists in spite of competence. An over-zealous concern to foster patient autonomy, at the expense of professional judgement and of seeking caregiver advice has potential to cause harm, for example through trying untested pharmaceutical interventions, allowing to continue driving or failing to facilitate access to aids and assistance when needed.

5.3 Summary

This study lends weight to research that previously identified a national and indeed global health system failure to adequately address co-morbid mental health problems occurring with neurological and other conditions with long term physical symptoms, in line with recommendations from the Royal College of Psychiatrists (2009) in the UK and the World Health Organisation (2006) report on the challenge to public health presented by neurological disorders. Specialist HD expertise is highly valued enabling timely input reflecting appreciation of needs at stages of the disease trajectory. Care, treatment and service provision need to adopt a flexible, integrated, interdisciplinary approach. Professionals need to value carer opinions and perspectives as well as their own judgement and balance them with the views of their

patients with HD. Support for centres providing specialist HD multi-disciplinary expertise is justified and methods of devolving their knowledge and skills to local services should be sought.

5.4 Relevance of the World Health Organisation (WHO) International Classification of Functioning, Disability and Health (ICF)

The ICF framework was referred to in the introduction to this thesis and in the methodology section as a conceptual model used as an aid to considering whether broader aspects of the health condition (in this case HD) were being addrerssed in this study. It was not used for classification.

In the methodology section it was suggested that if the concept mapping method was successful in presenting a comprehensive map of the the areas in which people with HD may need help towards living with the condition then the map would incorporate some statements relating to each component of the ICF model. Some of the resulting statements such as those that call for treatment for chorea and help with swallowing do relate to impairment of body function and structure. Limitations on activities are considered in statements that value help with sleeping, having sex, smoking and being able to drive. The range of statements forming the cluster labelled ‘social living’ closely relate to restrictions on participation. Statements about shop and town facilities, employers making reasonable adjustments to enable continued working and aids and adaptations incorporate environmental factors. The impact of personal factors is acknowledged in statements such as those concerned with genetic testing and the family implications of this, spending money while able, having a positive attitude and living for today. Regarding the factors influencing person’s functioning, *support and relationships* are addressed in statements concerned with family, friends and social network; attitudes are considered in statements about having community

understanding, visiting carers being flexible about the care they will give, being listened to as an adult and believed and respected; the *natural and human environment* is considered in statements to do with issues such as availability of manageable and appetising food, a care home environment being made to feel as a home should feel and having accessible town and shop facilities; *Services systems and policies* are addressed in statements about timely assessment for aids and adaptations, effective interagency working, and referral to a specialist HD centre; and *products and technology* are referred to in statements concerned with medication, aids and equipment and genetic testing.

Therefore the concept map derived from the study methodology has been found to embrace all components of the WHO ICF model adding to confidence that the method has successfully facilitated participants to be able to express views about a comprehensive range of areas of impact of the health condition (HD) where the various kinds of help were identified as being valued and needed.

The model conceptualises illness as a normal part of life experience: we will all be ill or impaired at some point and we can all experience disability to some degree (WHO 2001). The idea of 'disabled people' being distinct from the healthy population is seen as an untenable proposition as is the notion that health ends where disability begins (WHO 2001). It is a perspective that can serve well to illuminate the complexity of the caregiver / care-receiver relationship assumed in this study and in much of the HD literature. The hereditary nature of HD meant that some of those who participated as caregivers may well be positive for the HD mutation (whether or not confirmed through genetic testing) and may have already been affected by early symptoms which are often subtle and can include psychological and cognitive impairment long before motor signs are observable (Andersson et al 2012).

Caregivers tended to express annoyance that some health professionals believe patient reports rather than seek a caregiver perspective, arguing that the patient has impaired insight. But in some cases health professionals may be aware of possible impaired insight in the caregiver also. Williams et al (2012) highlight the complexity of caregiving roles given that multiple family members may be ill with HD.

The understanding within the ICF model that illness can be regarded as normal stresses the need to anticipate and plan for comorbidities among people with HD and this appears to explain the high priority given particularly by caregivers of doing so through timeliness in care delivery. The flexibility required by participants reflects the fluctuating state of health that the ICF model recognises. Health professionals need to remain aware that their time spent with the patient though critical (as opportunities to intervene and compensate for function and limit disability) is brief compared to the time the family spend trying to manage the impact of the condition. The patient presentation during consultations cannot be regarded as reliable and consistent; difficulties the family may face during periods of lower functioning must be taken into account in decision-making.

The finding that living with HD can be helped if provision of treatment, care and services is timely and flexible and that even more importantly the need for expert assessment and treatment for mental health conditions is appreciated strongly suggests a view among participants that the course and impact of the condition do not have to follow a trajectory of increasing despair. Instead this representative group established that the conclusion they draw from their experience is that the impact can be modified and managed despite relentless pathophysiological deterioration. Physiological changes alone are not held to determine the experience of living with HD- this has at least as much to do with organisation of health and

social care and welfare systems in our society. Whitehead and Dahlgren (2006) suggest that where differences in health are systematic and socially produced they are unfair and unjust. A lack of provision of psychiatric services for people with HD because the disease is considered by many psychiatrists to be a purely organic disorder is highlighted by Kenny and Wilson (2012), who indicate that people without HD would have easier access to assessment and treatment than people with HD presenting with the same psychiatric symptoms. This implies that people with HD suffer prolonged untreated psychiatric symptoms due to social inequity and injustice as described by Whitehead and Dahlgren (2006).

The model of the main determinants of health developed by Dahlgren and Whitehead (1991) illustrates how general socio-economic, cultural and environmental living and working conditions such as the work environment, education, unemployment and housing impact on the wellbeing of people living within their social networks. The model suggests that while individual factors such as lifestyle and constitution (in this case living with HD) are important determinants of personal health, policies and strategies targeted solely at the individual level will fail to affect important influences. Health care services are among the higher level determining factors that must be addressed in efforts to improve health for individuals (Dahlgren and Whitehead 1991).

The key findings of this study then can be considered to highlight inequality through the failure of the health care system to provide services to this minority group that are far more accessible to most members of society who need them.

5.5 Conclusions

Contribution to knowledge

The literature review in Chapter Two identified the need to understand patient, caregiver and health professional perspectives about what helps people to live with HD as a gap in the body of knowledge. The study has addressed that gap and has resulted in the first concept map that visually presents the way that an inclusive participant group conceptualise factors they consider to be helpful and how these factors are prioritised. The graphic representation facilitates explanation of the results to health professionals, researchers, voluntary organisations and HD family support groups so that decision-making about the kind of help to be sought and provided in future may be better informed.

Community and institutional contexts are accounted for. The study has shown that expert assessment and treatment for co-morbid mental health problems occurring with HD, access to professionals with HD expertise, timeliness and flexibility in the organisation and provision of care and services are regarded as the most important priority areas needing to be addressed. This has important implications for the current emphasis of research for effective treatments which is currently disproportionately focussed on the movement disorder.

The need for well-integrated interdisciplinary and interagency working that adopts a family-centred approach is also highlighted. There needs to be recognition of frustration felt by caregivers some of whom are also at risk of or have HD who are expected to provide ongoing care and often are left feeling excluded from discussions about what the patient needs. Professionals need to view principles of confidentiality as being in place to protect the privacy and dignity of patients rather

than to act as an obstacle to involving caregivers in joint planning to improve the quality of life of people with HD and their families. The importance of planning ahead to avoid crises rather than responding reactively is stressed and professionals must be aware that problems with insight and psychological defence mechanisms may prevent people with HD and some caregivers also from being able to initiate plans and ask for what they will need until it becomes too late. Therefore professionals must be prepared to proactively engage with the families to build a plan while working to enable those who cope by 'living for today' to maximise their potential physically, psychologically and socially in the present.

It is important to note that participants from all subgroups looked to healthcare systems to provide what is helpful and to note the hopes expressed by each regarding the kind of help most needed. For caregivers mainly this is expert care, aids, equipment and facilities; for people with HD this is effective medical treatment for the many aspects of their condition and support to carry on living independently for as long as possible and for professionals this is to strive to deliver on these hopes of patients and family caregivers.

Framework for dissemination

The non-neutral stance and the role of the primary investigator as an advocate for people with HD and family members have been acknowledged earlier (3.2). A background of involvement with support networks and care home settings and the position of a nurse educator have in the past served to facilitate activity aimed at contributing to improving care, treatment and services for this group. The findings of this study will now add confidence to making assertions about where help should be focussed on behalf of the stakeholders.

Responsibility for ensuring the findings of this study are disseminated effectively to maximise potential to benefit the HD community is acknowledged and regarded seriously. People with HD, family caregivers and health professionals have been generous with their time, knowledge and views, and have courageously engaged in sharing emotionally charged personal accounts of their experiences to illustrate and contextualise the statements they arrived at. Their trust that these efforts will make some difference and their entitlement to take the opportunity to express points of view and be heard will be respected through the implementation of an eight-point dissemination plan.

Support group meetings

Engagement with support groups for people with HD and family caregivers and groups of participating health care professionals has been ongoing since the planning stages and throughout the PhD process. The principal investigator met with the research steering group (HDA branch members) monthly during the year post data collection until the group disbanded due to practical difficulties relating to changed circumstances and has presented the preliminary results to each member.

Contact with all participating groups has been maintained and the principal investigator has visited each to update on progress at least once since data collection and has visited some of the groups several times. In May 2013 a programme of presenting findings to groups across England and Scotland will commence and this will include groups to which study participants are members. It is anticipated that nine engagements will be spread over the next 18 months.

People with HD, family caregivers and health professionals value these presentations as they are interactive and tend to reaffirm their shared sense of what is most important adding to their confidence in working to access what is needed.

European Huntington's Disease Network (EHDN) / International Huntington's Association (IHA) Involvement

Having frequently attended and previously presented at both of these international fora for researchers, health and social care professionals and families affected by HD, and as a member of two EHDN working groups (quality of life and advanced care working groups) the principal investigator will seek to utilise the platforms to present the study findings and initiate discussion about how to make service planners, health professionals and researchers aware of the highest priorities identified by the participants.

As a result of involvement with the EHDN the principal investigator has already taken part in a steering group supported by DeNDRoN and the HDA tasked with identifying priorities for HD researchers and preliminary findings presented to this group have influenced the strategy for working to establish areas for future research.

International Neuroscience Nursing Presentations

The principal investigator has presented aspects of this study including the design rationale, the development of the research question, the concept mapping method and key preliminary findings at neuroscience nursing conferences and symposia in Canada; Sweden; Belgium and England during progress. In the autumn of 2013 the findings will be presented at the World Federation of Neuroscience Nurses Congress in Gifu, Japan. Although primarily a nursing forum, this organisation is open to Allied Health Professionals and provides an opening to inform professionals with

broader neurological expertise about what is important to people with HD. In doing so a contribution towards improving HD expertise among generic neurological health experts as called for in the third highest priority cluster in this study.

Specialist Huntington's Facility engagements

One engagement to present the findings to staff at a large specialist HD care facility is arranged for autumn 2013 and the principal investigator will work to secure invitations to similar venues to do the same.

Elderly Care Facilities, Community Care team and Nursing Homes

As a link lecturer to various elderly care facilities in both hospital and nursing home settings and to a community care team where staff members occasionally encounter people with Huntington's disease the principal investigator frequently hosts seminars and will be able to present the findings of this study informally. Arguably the need to present these findings among staff who only occasionally encounter clients affected by the condition is even greater than among staff at a specialist HD centre where there is more likely to be a reasonable HD knowledge base.

Pre and Post Registration Nurse Education

As a lecturer in nursing and module leader for part of the second year undergraduate programme titled 'Long Term Conditions' the principal investigator has been able to ensure students learn about care for people with neurological disorders including Huntington's disease, Parkinson's disease, Multiple Sclerosis, Stroke and Alzheimer's disease. The students are given scenarios to explore in 'Enquiry-Based Learning', and the material provided to students is influenced by the results of this study.

Similarly the results are relevant to and will be integrated into a number of post registration nursing programmes that the principal investigator teaches on.

Journal Publications

Manuscripts will be submitted to peer reviewed journals including *Movement Disorders, Journal of Clinical Nursing, Journal of Advanced Nursing,* and *Journal of Huntington's Disease* for consideration. Papers to be based on the thesis will be:

- What helps people with Huntington's disease live with their condition?
- A review of perceived experience and responses to the challenges of Huntington's disease.
- Living with a neurological disorder: are the challenges generic or disease-specific?
- Concept Mapping: a novel method for nursing research

Involvement in Future Research Activity

At the time of completing this thesis the principal investigator has been appointed to a new post within the University of East Anglia School of Nursing Sciences . This will involve teaching and a supervised role as a member of an experienced team dedicated to securing research grant applications and undertaking high quality international research. The team has strong links with large private sector care provider organisations and a successful track record of research relating to neurological disorders and will provide a supportive learning environment fostering further development and opportunities to continue to engage in Huntington's disease-related research.

Limitations

People with HD and family carer participants were mainly Huntington's Disease Association (HDA) branch meeting attendees. They cannot be claimed to be representative of affected people and caregivers who do not have involvement with a supportive network such as the HDA.

The software programme (Ariadne®) used for analysis is less widely used than that designed and marketed by the developers of the concept mapping method (Concept Systems®, Kane and Trochim, 2007). However in an unpublished paper (in progress) a member of our own research team (Flaherty 2012) has identified over 240 health-related published concept mapping studies and a substantial minority use Ariadne® and found the software to be appropriate for concept mapping analysis (e.g. van Bon-Martens et al, 2011, Minkmann et al 2009). Ideally both software packages would have been used to analyse the data to check that results were similar but this was not possible due to financial restrictions.

Because of the impact on cognition of their advanced disease none of the participants who have HD, living in a nursing home were considered to have the capacity to understand and be able to carry out the prioritising or clustering tasks despite their valuable contribution to the brainstorming process. Therefore the attempt to compare a nursing home-based perspective to that of community-based participants only accounts for views of caregivers and health professionals. Clearly had it been possible such a comparison should have also represented views from people with HD.

Personal Reflections

While I have tried to maintain objectivity the influence of my own experience of professional involvement with people with HD during in choosing to study the topic of this thesis is acknowledged. To attempt to consider the nature of this influence I have reflected on the contexts of my past enagement: as a staff nurse in a specialised nursing home, as a guest at homes of numerous families affected by HD in a number of countries while on study tours and as a clinical nurse tutor at specialist care homes.

Using a structured model (Gibbs 1988) I carried out numerous personal reflections on this involvement over the fifteen years prior to commencing this study and finally combined my conclusions about what I felt I had learned from nursing this patient group as a staff nurse, nurse manager and clinical nurse tutor at various care home settings into a book which was distributed via the Huntington's Disease Association and the Scottish Huntington's Association mainly to affected families and to care home staff (Smith 2005) and later incorporated some of the conclusions into a further book (Smith 2011). I converted many of my reflections on anecdotes from families I stayed with in the UK, USA, Australia and Europe, into songs that I am frequently invited to perform at HD family meetings in many of those countries (Smith 2007).

This leads me to realise that I have been seeking to understand what helps people live with HD and to appreciate the perspectives of affected family members for many years prior to formulating the question for my PhD studies. In the books I drew together tips on how to give care that came from working with colleagues and observing what they did that seemed to be helpful. In the songs I presented what I

understood family members to be trying to convey about their experiences of living with HD.

Although I have acknowledged a non-neutral stance – clearly I had formulated some views on the topic ahead of undertaking the study- I am aware that I have been driven to investigate by a persistent sense of inadequacy that I have attributed to having no idea of what really helps people with HD or of what we professionals should regard as priorities when we seek to be helpful. I did not anticipate that addressing mental health problems would feature so highly in the results, but as I skim through my pre-study writings, although not specifically stated, I can see that the importance of flexibility and timeliness in care and service provision was appreciated and implied.

It would be unfortunate to carry out a study and fail to learn enough to be able to identify some aspects that with hindsight could have been improved on.

Design

The involvement of 40 members of the Huntington's Disease Association (caregivers and people with HD), and of health professionals and researchers within the supervision team and steering group, in critiquing and verifying the design and proposal for the ethics committee was a positive aspect, but a more diverse and representative consultation could have been achieved. Attendees at European Huntington's Association and the European Huntington's Disease Network include people with HD, family caregivers, health professionals and researchers with HD expertise. It is likely that a substantial number of volunteers from these groups would be keen to take part in sharing ideas to ensure international perspectives were accommodated. I am particularly pleased that the concept mapping approach

generated subjective opinions and allowed for quantitative analysis of how these were prioritised and conceptualised, and would be unlikely to have considered an alternative method. But ideas about how to recruit, particularly how to include people without involvement in support groups might be an example of an area where greater diversity of opinion regarding design detail could have been beneficial.

Recruitment

The failure to recruit sufficient numbers of participants for the study by the originally planned method of access via a clinic database and the need to resort to recruiting from HDA branches may be informative with regard to ways in which many affected individuals and family members tend to deal with the condition. There appears to be very little evidence to inform an explanation for this but I can lend anecdotal support to a small qualitative study by Lowit and Teijlingen (2005) involving 10 HD family caregivers. The study found that whereas people with other serious health conditions and their caregivers value support meetings, for many people with HD and family members the advantages of avoiding confrontation with the future impact of HD by meeting others perhaps with more advanced disease exceed any benefits of feeling supported within groups.

When potential recruits from the clinic database declined to take part in the study no reason was asked for but the wish to avoid having to think about what help may be needed in future and particularly to avoid meeting anyone with more obvious symptoms than themselves was frequently expressed, in keeping with the findings of Lowit and Teijlingen (2005). The consequent low representation of non-members of support groups in my own study sample is acknowledged and there appears to be justification for considering further qualitative studies in this area that do not adopt a

group-orientated design. That said, there is no evidence to indicate that the HD support group members who took part in my own study did not share views held by non-group members. Some participants did take part individually and on reflection I feel confident that efforts to limit this potential for recruitment bias resulted in a meaningful representation of what is felt by stakeholders to help people live with HD.

Having touched on the issue of recruitment, inclusion of participants from various European countries, and perhaps beyond would also have been achievable and could add to confidence regarding the relevance of the results beyond this country. Near to the end of my data collection activity, members of a large support group in Scotland expressed an interest in participating. I had to decline because my ethics permission did not extend into Scotland. It would have been possible to obtain permission in the region but time would not allow. With hindsight I should have anticipated this and sought Scottish permission earlier.

Overall lesson learned

I would consider utilising an electronic means of allowing people to enter their prioritising and clustering data. Many people asked for this, and many professionals in particular would have been happier to complete both tasks if this had been an option.

I have learned that with any large project it is important to start writing up early, rather than rely on notes at a later date. I found that it is useful to read exemplar theses early on, and get a feel for what it should look like. I have gained greater confidence that by carrying out research I am not perceived as a nuisance. It is a worthwhile contribution. I shouldn't have feared so much all the time that people

would see it as such an intrusion. Feedback about how people found the activities therapeutic should be more readily believed. That said, I would still prefer to err on the side of caution, respecting privacy and dignity, but when they indicate being keen to take part, I should relax a little more and believe them.

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Appendices

Appendix 3.1 Authorisation documents including Research Ethics Approval for study and amendment, University approval and insurance documents.

NHS

National Research Ethics Service

Essex 2 Research Ethics Committee
Terminus House
9th Floor
The High
Harlow
Essex
CM20 1XA

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25 September 2009

Mr Steve Smith
Lecturer, Nursing.
University of East Anglia.
School of Nursing and Midwifery
Edith Cavell Building
University of East Anglia, Norwich.
NR4 7TJ

Dear Mr Smith

Study Title:	**A Concept Map of What Helps People With Huntington's Disease Manage Their Condition: Comparing Perspectives of People With HD, Carers, and Health Professionals.**
REC reference number:	**09/H0302/87**
Protocol number:	**Version 4.0**

Thank you for your letter of 09 September 2009, responding to the Committee's request for further information on the above research and submitting revised documentation.

The further information was considered in correspondence by a sub-committee of the REC [at a meeting held on [date].] A list of the sub-committee members is attached.

Confirmation of ethical opinion

On behalf of the Committee, I am pleased to confirm a favourable ethical opinion for the above research on the basis described in the application form, protocol and supporting documentation [as revised], subject to the conditions specified below.

Mental Capacity Act 2005

I confirm that the committee has approved this research project for the purposes of the Mental Capacity Act 2005. The committee is satisfied that the requirements of section 31 of the Act will be met in relation to research carried out as part of this project on, or in relation to, a person who lacks capacity to consent to taking part in the project.

Ethical review of research sites

The favourable opinion applies to all NHS sites taking part in the study, subject to management permission being obtained from the NHS/HSC R&D office prior to the start of the study (see "Conditions of the favourable opinion" below).

Conditions of the favourable opinion

The favourable opinion is subject to the following conditions being met prior to the start of the study.

1. Management permission or approval must be obtained from each host organisation prior to the start of the study at the site concerned.

2. For NHS research sites only, management permission for research ("R&D approval") should be obtained from the relevant care organisation(s) in accordance with NHS research governance arrangements. Guidance on applying for NHS permission for research is available in the Integrated Research Application System or at http://www.rdforum.nhs.uk. *Where the only involvement of the NHS organisation is as a Participant Identification Centre, management permission for research is not required but the R&D office should be notified of the study. Guidance should be sought from the R&D office where necessary.*

3. Sponsors are not required to notify the Committee of approvals from host organisations.

Other conditions specified by the REC

1. The version number and date on the Information for Health Professionals is incorrect and must be amended to version 5.0 dated 07.09.09. Final versions of documents should be provided to the committee for information,

It is the responsibility of the sponsor to ensure that all the conditions are complied with before the start of the study or its initiation at a particular site (as applicable).

Approved documents

The final list of documents reviewed and approved by the Committee is as follows:

Document	*Version*	*Date*
Lone Working Protocol	Version 1.0	22 June 2009
Information for Consultees	Version 2.0	22 June 2009
Consultee confirmation willing to take part	Version 2.0	22 June 2009
Consultee Reply Slip	Version 2.0	22 June 2009
Supervisor CV Dr G D'Cruz		
Participant Consent Form: People with severe HD	Version 2.0	22 June 2009
Participant Consent Form: People with HD	Version 2.0	22 June 2009
Participant Consent Form: Carers of people with HD	Version 2.0	22 June 2009
Participant Consent Form: Professional	Version 2.0	22 June 2009
GP/Consultant Information Sheets	Version 2.0	22 June 2009
Covering Letter		23 June 2009
Investigator CV		22 June 2009
REC application	Version 2.0	24 June 2009
Letter of invitation to participant	Version 1.0	22 June 2009
Letter of invitation to participant	Version 1.0	22 June 2009
Letter of invitation to participant	Version 1.0	22 June 2009
UEA liability confirmation letter		24 June 2009
Letter of invitation to participant	Version 1.0	22 June 2009
Protocol	Version 4.0	05 September 2009

Participant Information Sheet: Carers	Version 5.0	07 September 2009
Participant Information Sheet: People with severe HD	Version 5.0	07 September 2009
Participant Information Sheet: People with HD	Version 5.0	07 September 2009
Participant Information Sheet: Health Professionals	Version 4.0	22 June 2009
Letter from Katherine Deane		
Response to Request for Further Information		09 September 2009

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

After ethical review

Now that you have completed the application process please visit the National Research Ethics Service website > After Review

You are invited to give your view of the service that you have received from the National Research Ethics Service and the application procedure. If you wish to make your views known please use the feedback form available on the website.

The attached document *"After ethical review – guidance for researchers"* gives detailed guidance on reporting requirements for studies with a favourable opinion, including:

- Notifying substantial amendments
- Adding new sites and investigators
- Progress and safety reports
- Notifying the end of the study

The NRES website also provides guidance on these topics, which is updated in the light of changes in reporting requirements or procedures.

We would also like to inform you that we consult regularly with stakeholders to improve our service. If you would like to join our Reference Group please email referencegroup@nres.npsa.nhs.uk.

09/H0302/87	**Please quote this number on all correspondence**

Yours sincerely

pp

Mr Jon Gould
Chair
Email: suzanne.emerton@eoe.nhs.uk

Enclosures: List of names and professions of members who were present at the meeting and those who submitted written comments [if final opinion was confirmed was given at a meeting]

"After ethical review – guidance for researchers"

NHS

National Research Ethics Service

Essex 2 Research Ethics Committee

Terminus House
9th Floor
The High
Harlow
Essex
CM20 1XA

Tel: 01279 419312
Fax: 01279 419246

13 November 2009

Mr Steve Smith
Lecturer, Nursing.
University of East Anglia.
School of Nursing and Midwifery
Edith Cavell Building
University of East Anglia, Norwich.
NR4 7TJ

Dear Mr Smith

Study title:	**A Concept Map of What Helps People With Huntington's Disease Manage Their Condition: Comparing Perspectives of People With HD, Carers, and Health Professionals.**
REC reference:	**09/H0302/87**
Amendment number:	**AM 01**
Amendment date:	**27 October 2009**

The above amendment was reviewed at the meeting of the Sub-Committee held on 06 November 2009by the Sub-Committee in correspondence.

Ethical opinion

The members of the Committee taking part in the review gave a favourable ethical opinion of the amendment on the basis described in the notice of amendment form and supporting documentation.

Approved documents

The documents reviewed and approved at the meeting were:

Document	Version	Date
Letter of invitation to participant	AM 01	27 October 2009
Participant Information Sheet: Carers	AM 01	27 October 2009
Participant Information Sheet: With HD	AM 01	27 October 2009
Protocol	AM 01	27 October 2009
Notice of Substantial Amendment (non-CTIMPs)	AM 01	27 October 2009
Letter of invitation to participant	AM 01	27 October 2009
Letter of invitation to participant	AM 01	27 October 2009

Membership of the Committee

The members of the Committee who took part in the review are listed on the attached sheet.

R&D approval

All investigators and research collaborators in the NHS should notify the R&D office for the relevant NHS care organisation of this amendment and check whether it affects R&D approval of the research.

Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

09/H0302/87:	**Please quote this number on all correspondence**

Yours sincerely

Suzanne Emerton

Suzanne Emerton
Committee Co-ordinator

E-mail: suzanne.emerton@eoe.nhs.uk

Enclosures: List of names and professions of members who took part in the review

Copy to: Ms Tracy Moulton
Research Contract Manager
University of East Anglia
Norwich
NR4 7TJ

Addenbrookes Hospital R & D Dept

University of East Anglia

The Registry
Research, Enterprise and Engagement Office

University of East Anglia
Norwich NR4 7TJ
England

Tel: +44 (0) 1603 456161
Direct: +44 (0) 1603 591574
Fax: +44 (0) 1603 591550
Email: reeo@uea.ac.uk
www.uea.ac.uk/reeo

TO WHOM IT MAY CONCERN

24 June 2009

Dear Sirs

Study: A Concept Map of What Helps People With Huntington's Disease Manage Their Condition: Comparing Perspectives of People With HD, Carers, and Health Professionals.
Chief Investigator: Steve Smith

This is to confirm that the University of East Anglia and Subsidiary Companies has arranged insurance cover with Zurich Municipal (Policy No. NHE-09AC01-0013) with limits of liability as detailed below. The Policy is due to expire on 31 May 2010 when we expect to renew on substantially similar terms:-

Employers Liability	- £25,000,000 per claim
Public/Products Liability	- £25,000,000 per claim
Professional Negligence	- £ 7,500,000 per claim

The cover is, of course, subject to the terms and conditions of the policy. If you require further details, please contact the undersigned.

Yours faithfully

T. Moulton

Tracy Moulton
Research Contracts Manager
Research, Enterprise and Engagement Office
University of East Anglia
Norwich NR4 7TJ

Tel: 01603 591482 / Fax: 01603 591550
Email: t.moulton@uea.ac.uk

Faculty of Health
Postgraduate Programmes Office

University of East Anglia
Norwich NR4 7TJ England

Telephone: 01603 456161

Direct dial: 01603 591258
Fax: 01603 593166
Email: foh.pgr@uea.ac.uk

30 July 2008

TO WHOM IT MAY CONCERN

Student Name:	Mr Stephen Smith
Registration Date:	1 April 2007
Student Registration Number:	3570363
Course:	MPhil/PhD
School:	School of Nursing & Midwifery

This is to confirm that Mr Smith is currently registered as a postgraduate student on the part-time MPhil/PhD research degree programme in the School of Nursing & Midwifery. His period of study runs from the 1 April 2007 to 31 March 2013.

If you require any further information, please contact me.

Yours sincerely

Gillian Potter
Senior Administrative Assistant
Faculty of Health
University of East Anglia
016903 591258

University of East Anglia

The Registry
Research, Enterprise and Engagement Office

University of East Anglia
Norwich NR4 7TJ
England

Tel: +44 (0) 1603 456161
Direct: +44 (0) 1603 591574
Fax: +44 (0) 1603 591550
Email: reeo@uea.ac.uk
www.uea.ac.uk/reeo

3 November 2009

Dear Mr Smith.

REC No. 09/H0302/87
A Concept Map of What Helps People With Huntington's Disease Manage Their Condition: Comparing Perspectives of People With HD, Carers, and Health Professionals.

I can confirm that the University of East Anglia will act as sponsors for this research project with respect to the UK Department of Health's Research Governance Framework for Health and Social Care.

Sponsorship is granted under the understanding that you as the Chief Investigator will conduct the research project in compliance with:

a. all relevant University and, if applicable, Trust policies and procedures (which can be found on the respective websites)
b. the UK's Department of Health's Research Governance Framework for Health and Social Care;

Yours sincerely

T. Moulton

Tracy Moulton
Research Contracts Manager
Research, Enterprise and Engagement Office
University of East Anglia
Norwich NR4 7TJ

Tel: 01603 591482 / Fax: 01603 591550
Email: t.moulton@uea.ac.uk

Appendix 3.2 Recruitment documents including invitation letters, information document, consent forms and reply slips.

3.2A: Letter of invitation to take part addressed to people with HD

University of Cambridge

Dr. Roger Barker, PhD, MRCP
Centre for Brain Repair
Forvie Site, Robinson Way
Cambridge, CB2 2PY
UK

Tel: +44 (0)1223 331184

Fax: +44 (0)1223 331174

Email: rab46@cam.ac.uk

Cambridge Centre for Brain Repair

Addenbrooke's NHS Trust

Date ----------

Name Address of Recipient Here

Dear ----------

You are invited to join in with some research about Huntington's disease (HD). If you would like to know what this research is about please read the documents that are with this letter. You may want help with reading them. You could ask a carer to help, or talk to your doctor (GP). You could ask them to help you decide if it's a good idea to join in with the research.

Also, do you have a 'main' carer? I mean, someone who helps you manage your HD, who would be the right person to talk about caring for you, in this study? If so, please would you give that person the enclosed 'Carer Pack'? Thank you.

In about a week, Steve Smith, the Chief Investigator for the study, will ring you to ask if you are interested in taking part. If you are, he will arrange to visit you at your home, to discuss what is involved. Just tell Steve if you don't want to take part, it is not a problem.

If you have any questions about the study, please contact **Mr Steve Smith, Lecturer University of East Anglia, Faculty of Health, School of Nursing and Midwifery, Edith Cavell Building, Norwich NR4 7TJ.** Or ring and leave a message on 01603 597022, or e mail steve.smith@uea.ac.uk . Steve will get back to you.

Very many thanks,

Yours Sincerely

Dr Roger Barker

Appendix 3.2 B: Letter of invitation to take part addressed to caregegivers for people with HD

University of Cambridge

Cambridge Centre for Brain Repair

Addenbrooke's NHS Trust

Dr. Roger Barker, PhD, MRCP
Centre for Brain Repair
Forvie Site, Robinson Way
Cambridge, CB2 2PY
UK

Tel: +44 (0)1223 331184

Fax: +44 (0)1223 331174

Email: rab46@cam.ac.uk

Date ----------

Name Address of Recipient Here

Dear ----------

You are invited to take part in a research project. It is to do with your role as a carer for someone who has Huntington's disease (HD), and has also been invited to take part. The person with HD was asked to give this 'carer pack' to the person they wish to be regarded as their carer for this study.

In about one week after this letter is posted, the Chief Investigator, Steve Smith, will ring you to ask if you are interested in taking part. If you are he will arrange to visit you to discuss what the study involves. Meanwhile, if you are interested, please read the enclosed information. There is no need to fill in or sign any forms yet.

If you wish to ask any questions about the project meanwhile, please contact: **Mr Steve Smith, Lecturer,**

University of East Anglia, Faculty of Health,

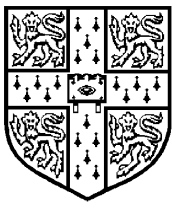
School of Nursing and Midwifery, Edith Cavell Building, Norwich NR4 7TJ. Or ring and leave a message on 01603 597022, or e mail steve.smith@uea.ac.uk . Steve will get back to you.

Very many thanks,

Yours Sincerely

Dr. Roger Barker.

Appendix 3.2 C Letter of invitation to take part addressed to Health Professionals



University of Cambridge

Cambridge Centre for Brain Repair

Addenbrooke's NHS Trust

Dr. Roger Barker, PhD, MRCP
Centre for Brain Repair
Forvie Site, Robinson Way
Cambridge, CB2 2PY
UK

Tel: +44 (0)1223 331184

Fax: +44 (0)1223 331174

Email: rab46@cam.ac.uk

Date ----------

Name Address of Recipient Here

Dear ----------

You are invited to take part in a research project related to your work as a Health-related Professional, providing or organising care and / or support for people with Huntington's disease. If you may be interested to know about the project, please read the enclosed information document.

About a week after sending you this, the Chief Investigator, Steve Smith, will ring you to ask whether you are interested in taking part in the study. If you are, he will make arrangements with you regarding taking consent and details of meetings to carry out the study.

If you would like to discuss the project or ask any questions about it, please contact:

Steve Smith, Lecturer, University of East Anglia, Faculty of Health, School of Nursing and Midwifery, Edith Cavell Building, Norwich NR4 7TJ. Or ring and leave a message on 01603 597022. or e mail steve.smith@uea.ac.uk . Steve will get back to you.

Very many thanks,

Yours Sincerely

Dr Roger Barker

Appendix 3.2D Information document for people with HD

Information Sheet For People who Have HD.

1. Title: **What helps me manage my Huntington's disease?**

2. **Invitation**

Would you like to join in with some research about Huntington's disease, or 'HD'? It would mean talking about your HD. Nothing would be done to you. A researcher called Steve who is a nurse would be asking you some questions.

You do not have to do this. Before you decide if you want to join in, it is important for you to understand what the research is for and what you would be doing.

Please take time to read the writing below, or get someone to help you or read it to you.

It is a good idea to talk to others about it if you're not sure whether to join in. You could speak to your carer or to your GP (doctor). Do get in touch with us if you want to know more about it.

Thank you for reading this.

3. **What is the research for?**

We hope to find out what helps people with Huntington's disease (HD) manage their HD. We want to know what people with HD and their carers and doctors and nurses and others who care think about this.

We would like to know if doctors and nurses and other professionals know what people with HD think is helpful. And we want to know how important these things are to you.

We think this will help people who care to be more helpful to people with HD in future.

4. **Why was I chosen?**

You were one of 13 people with HD chosen from a list kept at the Brain Repair Centre Clinic in Cambridge. 10 health professionals, were also chosen. And we want to enrol 10 carers, chosen by people with HD.

5. **Do I have to join in?**

It is up to you to decide if you want to join in. If you decide to join in you can change your mind at any time. It will not affect the care you receive. You don't have to give a reason if you stop joining in the research.

6. **What do I have to do?**

In about a week after sending you this information, the Chief Investigator (Steve) will ring you to see if you are interested to join in. If you don't want to, just tell Steve. It's not a problem. If you are interested, Steve will arrange to visit you to discuss the study, and make sure you understand what you would have to do.

If you agree to join in, Steve will ask you to sign the consent form and fill in the reply slip. You can keep a copy of each of these documents so that you can read them at any time.

If you take part, this will mean coming to two meetings. Steve will discuss with you how you will get there.

The first meeting will last two hours, with a break in the middle for refreshments. This meeting will be with about 7 other people who have HD. Steve and an assistant will be there to lead the meeting and help if you're not sure what to do. Your carer may be in the building, in case he or she is needed, but will not be in the meeting with you. The idea of this meeting is to get ideas only from people who have HD.

We will ask you all as a group, to agree on statements about things that you think help you to manage your HD.

The second meeting will also last two hours, with a break for refreshments. This time the other people with HD, and carers, and health professionals will all be there. This meeting will be about deciding which statements about what helps people to manage their HD are most important, and which are not so important.

On the day we will be there to help to make sure you understand how to do it. But there will be no 'right' or 'wrong' answers. We want your ideas.

There is one more thing we would like you to help with. After the meetings are finished Steve and the research team will work to make a report of the results of the study. We would like to send you a copy of a draft of this report, for you to comment on. You could point out anything you do or don't agree with. Then a final report will be written, and you will be given a copy. The Huntington's Disease Association will also receive a copy.

An important thing to note is that we will audio record the meetings. This is to help us remember what was said as we write about the study. It will help us if we are not sure what any written statements mean. After the report is written the tape will be destroyed. H

7. **Are there any risks to me?**

Talking about how HD has affected you could upset you and make you think about some things you like to forget.

8. **Could joining in the study do me some good?**

On the other hand, it might feel good to talk about things that help you to manage. You might feel good that this will help people in future, to know what helps people with HD.

But, the research will not help you personally.

9. **Will my taking part in this study be kept confidential?**

Steve will keep the forms you sign with your name and other details locked safely. He may publish some of your ideas but will not tell anyone your personal details. Steve will tape record what you say to remind him later. The tape will be kept locked safely and will be destroyed when the research is finished.

The others in the room will be asked to keep what you say confidential. But we cannot guarantee they will. You do not have to say anything that you don't want others to know.

10. **What will happen to the results of the research study?**

We will try to publish results in research journals to do with health. Also, we will give the Huntington's Disease Association a report, to print in the newsletter if they wish.

The chief investigator (Steve) will talk about the study at meetings about HD.

11. Who is organising and funding the research?

This study is part of a PhD project for the chief investigator (Steve), and is paid for by the University of East Anglia, School of Nursing Midwifery.

12. Who has reviewed the study?

The study has been reviewed by experienced health researchers at the University of East Anglia. It has been approved by the Local Research Ethics Committee. It will be monitored by a research team including experienced researchers, a person with HD, and a carer, and a Family Care Advisor from the Huntington's Disease Association.

13. Contact for Further Information

Very many thanks for taking part in this study if you do decide to. If you wish to discuss it further or ask any questions about it, please contact the chief investigator:

Steve Smith
University of East Anglia,
Faculty of Health,
School of Nursing and Midwifery
Edith Cavell Building
Norwich
NR4 7TJ.
Tel: 01603 59 7022 (leave a message if not there and Steve will get back to you)
Or e mail: steve.smith@uea.ac.uk

Declaration:

I have read and understood this information sheet.

Signed _____ Name (Print)

_____

Date _____

OR:

Declaration:

My carer has read this information sheet to me and I understand it.

Signed _____

Name (Print) _____ Date _____

Please Note: You should sign and return one copy of this information sheet and a consent form, and keep a 2nd copy of each.

Reply Slip

If you have decided that you wish to take part in the study: 'What Helps Me Manage My Huntington's Disease?', please complete this reply slip or ask you carer to fill it in for you, with YOUR answers, and hand it to Steve Smith with the consent form, signed and dated, and the information sheet, also signed and dated (one copy only of each document).

Full Name _____ Date of Birth _____

Address_____

_____

Phone _____

How long is it since you were first affected by the symptoms of HD?

_____

Signed _____ Date _____

Appendix 3.2 E Information document for caregivers for people with HD

Information Sheet For Carers of People With HD.

1. Title: **What helps me manage my Huntington's disease?**

2. **Invitation**

Would you like to join in with some research about Huntington's disease, or 'HD'? It would mean talking about your experience of caring for a person who has HD. Nothing would be done to you. A researcher called Steve who is a nurse would be asking you some questions.

You do not have to do this. Before you decide if you want to join in, it is important for you to understand what the research is for and what you would be doing.

Please take time to read the writing below, or get someone to help you or read it to you.

It is a good idea to talk to others about it if you're not sure whether to join in. You could speak to your GP (doctor) about it. Do get in touch with us if you want to know more about it.

Thank you for reading this.

3. **What is the research for?**

We hope to find out what helps people with Huntington's disease (HD) manage their HD. We want to know what people with HD and their carers and doctors and nurses and others who care think about this.

We would like to know if doctors and nurses and other professionals know what people with HD and their carers think is helpful. And we want to know how important these things are to you.

We think this will help people who care to be more helpful to people with HD in future.

4. **Why was I chosen?**

You were chosen by the person you care for who has HD. He or she was sent an invitation and a pack for carers, and was asked to give the 'carer pack' to the person they feel can best act as their main carer for this study. The person you care for with HD is one of 13 people with HD chosen from a list kept at the Brain Repair Centre Clinic in Cambridge. 10 health professionals, were also chosen.

5. **Do I have to join in?**

It is up to you to decide if you want to join in. If you decide to join in you can change your mind at any time. It will not affect the care you receive. You don't have to give a reason if you stop joining in the research.

6. **What do I have to do?**

In about a week after sending the person you care for, who has HD this information, the Chief Investigator (Steve) will ring to see if he or she, and if you are interested to join in. If you don't want to, just tell Steve. It's not a problem. If you are both interested, Steve will arrange to visit you to discuss the study, and make sure you understand what you would have to do.

If you agree to join in, Steve will ask you to sign the consent form and fill in the reply slip. You can keep a copy of each of these documents so that you can read them at any time.

If you take part, this will mean coming to two meetings. Steve will discuss with you how you will get there.

The first meeting will last two hours, with a break in the middle for refreshments. This meeting will be with about 9 other people who care for someone who has HD. Steve and an assistant will be there to lead the meeting and help if you're not sure what to do.

We will ask you all as a group, to agree on statements about things that you think help people to manage their HD.

The second meeting will also last two hours, with a break for refreshments. This time, people with HD, and health professionals will all be there as well as carers. This meeting will be about deciding which statements about what helps people to manage their HD are most important, and which are not so important.

On the day we will be there to help to make sure you understand how to do it. But there will be no 'right' or 'wrong' answers. We want your ideas.

There is one more thing we would like you to help with. After the meetings are finished Steve and the research team will work to make a report of the results of the study. We would like to send you a copy of a draft of this report, for you to comment on. You could point out anything you do or don't agree with. Then a final report will be written, and you will be given a copy. The Huntington's Disease Association will also get a copy.

An important thing to note is that we will audio record the meetings. This is to help us remember what was said as we write about the study. It will help us if we are not sure what any written statements mean. After the report is written the tape will be destroyed.

7. **Are there any risks to me?**

Talking about how caring for someone who has HD has affected you could upset you and make you think about some things you like to forget.

8. **Could joining in the study do me some good?**

On the other hand, it might feel good to talk about things that help the person you care for to manage. And what helps you. You might feel good that this will help people in future, to know what helps people with HD.

But, the research will not help you personally.

9. **Will my taking part in this study be kept confidential?**

Steve will keep the forms you sign with your name and other details locked safely. He may publish some of your ideas but will not tell anyone your personal details. Steve will tape record what you say to remind him later. The tape will be kept locked safely and will be destroyed when the research is finished.

The others in the room will be asked to keep what you say confidential. But we cannot guarantee they will. You do not have to say anything that you don't want others to know.

10. **What will happen to the results of the research study?**

We will try to publish results in research journals to do with health. Also, we will give the Huntington's Disease Association a report, to print in the newsletter if they wish.

The chief investigator (Steve) will talk about the study at meetings about HD.

11. Who is organising and funding the research?

This study is part of a PhD project for the chief investigator (Steve), and is paid for by the University of East Anglia, School of Nursing Midwifery.

12. Who has reviewed the study?

The study has been reviewed by experienced health researchers at the University of East Anglia. It has been approved by the Local Research Ethics Committee. It will be monitored by a research team including experienced researchers, a person with HD, and a carer, and a Family Care Advisor from the Huntington's Disease Association.

13. Contact for Further Information

Very many thanks for taking part in this study if you do decide to. If you wish to discuss it further or ask any questions about it, please contact the chief investigator:

Steve Smith
University of East Anglia,
Faculty of Health,
School of Nursing and Midwifery
Edith Cavell Building
Norwich
NR4 7TJ.

Tel: 01603 59 7022 (leave a message if not there and Steve will get back to you)

Or e mail: steve.smith@uea.ac.uk

Declaration:

I have read and understood this information sheet.

Signed _____ Name (Print)

_____

Date _____

Carer information sheet version 4, 11 06 09.

Please Note: You should sign and return one copy of this information sheet and a consent form, and keep a 2nd copy of each.

Reply Slip

If you have decided that you wish to take part in the study: 'What Helps Me Manage My Huntington's Disease?', please complete this slip and send it to Steve Smith with the consent form, signed and dated, and the information sheet, also signed and dated (one copy only of each document).

Full Name _____

Date of Birth _____

Gender Male Female (please delete as applicable).

Address_____

_____

How long have you cared for the person with HD taking part in this study?

_____

If you have cared for others with HD, how many years have you spent caring for a person with HD in total? _____

What is your relationship to the person with HD that you are caring for?

_____

Name Address and phone number of your GP.

_____

Phone number or e mail address for the researcher (Steve) to contact you on, if needed, to discuss arrangements for the meetings:

_____

Signed _____ Date _____

Appendix 3.2 F Information document for Health Professionals

Information Sheet For Health Professionals

1. Title: **What helps me manage my Huntington's disease?**

2. **Invitation**

You are being invited to take part in a research study. Before you decide whether to accept, it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully and discuss it with others if you wish. Ask us if there is anything that is not clear or if you would like more information.

Thank you for reading this.

3. **What is the purpose of the study?**

The aim of this study is to determine what helps people with Huntington's disease (HD) manage their condition, from the perspectives of people with HD, their carers and of health professionals- that is, qualified health-related professionals such as nurses, doctors, physiotherapists, and speech and language therapists, and social workers.

The study will examine similarities and differences in the views of each of these three groups regarding what can help people to manage their HD, and the level of priority accorded to factors that can help.

It is anticipated that the knowledge resulting from the study will be useful to health professionals, carers and people with HD in planning how they can work together to make it easier to manage life with HD.

4. **Why have I been chosen?**

You were among 10 health and social care professionals selected from attendance lists at professional meetings concerned with care for people with Huntington's disease (HD). The selection was by 'purposive' sampling- this means ensuring representation from a range of professional backgrounds, and convenience- geographically close to limit potential barriers to meeting. 13 people who are diagnosed with HD (including 5 with advanced HD) and 10 carers have also been recruited to take part.

5. **Do I have to take part?**

It is up to you to decide whether or not to take part. In about one week after sending you this invitation, the Chief Investigator, Steve Smith, will ring you to see if you are interested and answer any questions you may have about the study. If you do decide to take part you should sign one copy of this information sheet, and reply slip giving your contact details, and sign a copy of the consent form, and return these signed documents to Steve Smith in the pre-paid envelope.

You should keep the other copies of each document. If you decide to take part you are still free to withdraw at any time and without giving a reason.

6. What do I have to do?

If you agree to take part, and return the completed and signed consent form and reply slip to the chief investigator in the enclosed stamped addressed envelope, then the chief investigator will contact you to make arrangements regarding the details of two meetings with other health professionals. The meetings are each expected to last about two hours including a break with light refreshment provided. If this commitment seems too demanding, it is possible to arrange to do the second activity individually, from your home or work base.

In the first meeting, the group are asked to try to agree on statements that identify some of things that can be most helpful towards enabling people with HD to manage their condition. In the first meeting you will be with other health professionals, and not with people with HD or their carers.

The second meeting can be attended by all three groups together- people with HD, carers and Health professionals, but, as said, above, you can carry out the second meeting activities on your own, at a time and location you prefer. This second session involves deciding an order of priority for the statements and then grouping them into themes, or 'clusters'.

Reasonable travel expenses will be reimbursed.

11. What are the possible disadvantages and risks of taking part?

A risk assessment process has highlighted the potential situation in which the chief investigator becomes aware of unprofessional practice, potentially harmful to a patient with HD. In such circumstances the chief investigator would be duty bound to report the matter to the appropriate authority, after notifying the health professional concerned.

12. What are the possible benefits of taking part?

On the other hand, you may benefit by getting to discuss some difficult issues with others who have worked to help patients with HD or their caregivers to manage them. You may also feel a benefit from the satisfaction of taking part in research that is aimed at improving understanding of HD and how it affects individuals, carers and

families, and at bringing the aims of interventions by health professionals more in line with the needs of the people they work to help. With an increasing emphasis on evaluating performance in terms of patient reported outcomes (PRO's), greater understanding of any differences between patient, caregiver and clinician perspectives may benefit your future work in this area.

That said, it should be emphasised that there is no claim to offer a direct definite benefit to you personally.

16. **Will my taking part in this study be kept confidential?**

Clearly, the members of the group will get to know each other to some extent, and will know what you have said during discussions. While the chief investigator will emphasise to the group, the importance of keeping any personal information discussed confidential, he cannot guarantee that all members will do so. You should feel under no pressure to disclose any personal information that you do not wish to share with the group, bearing in mind that someone may repeat it.

Otherwise, yes, your taking part will be kept confidential. Written records of your personal identifying details held will be kept on computer with password security, and on a document also protected by further password security.

The group discussions will be tape recorded for analysis. The tape will be kept under secure conditions until analysed and then will be destroyed. The security arrangements have been approved by the regional ethics committee.

Regarding publication of results, no personal identifying details of participants will be published.

17. **What will happen to the results of the research study?**

The aim is that results will be published in research journals related to health, for example, Neurology, or International Journal of Quality of Life Research, or the Journal of Clinical Nursing, or Journal of Advanced Nursing. Also, a report will be given to the Huntington's Disease Association, for publication in the newsletter and as they deem appropriate. Participants will be invited to comment on a draft of the report before it is finalised, and will be given a copy of the final report.

The chief investigator frequently presents to professional and family groups associated with HD, in the UK and abroad and will present a summary wherever appropriate.

18. **Who is organising and funding the research?**

This study is being carried out as a PhD project for the chief investigator, and is supported and funded by the University of East Anglia, School of Nursing Midwifery, within the Faculty of Health.

19. Who has reviewed the study?

The study has been overseen and reviewed by a research team including three experienced health researchers (supervisors, including health professionals) at the University of East Anglia, Faculty of Health, a person with HD, a carer and a representative from the Huntington's Disease Association. It has also been approved by the Local Research Ethics Committee.

20. Contact for Further Information

Very many thanks for taking part in this study if you do decide to. If you wish to discuss it further or ask any questions about it, please contact the chief investigator:

Steve Smith
University of East Anglia,
Faculty of Health,
School of Nursing and Midwifery
Edith Cavell Building
Norwich
NR4 7TJ.
Tel: 01603 59 7022 (leave a message if not there and Steve will get back to you)
Or e mail: steve.smith@uea.ac.uk

Reply Slip

Declaration:

I have read and understood this information sheet.

Signed _____ Name (Print)
_____

Date _____

Please Note: You should sign and return one copy of this information sheet and a consent form, and keep a 2nd copy of each.

Please complete the following:

Name of profession and position _____

Address_____
_____

Date of Birth _____ Gender (tick as applicable) M □ F □

How long (in years) have you had professional experience of involvement with people who have Huntington's disease? ----------

How many patients with HD have you been professionally involved with in the past two years? _____

Daytime phone number _____

E mail address _____

Appendix 3.2 G Consent form for people with HD

Consent Form

For people with Huntington's Disease

Title: **What Helps Me manage My Huntington's Disease?**

Name of Researcher: Steve Smith

Please tick to confirm

I confirm that I have read and understand the information sheet for the research project called 'What Helps Me manage My Huntington's Disease?'

□

I have had a chance to think about it and ask any questions. I am sure that I know enough about it to help me decide about joining in.

□

I understand that the sessions will be recorded, and after the researchers analyse the statements with the aid of the recording, the tape will be destroyed. □

I know that I don't have to do this, It is my own choice. If I start joining in with the research I know that I can stop if want to at any time. I will still be cared for in the same way, whether I join in or not.

□

I agree to take part in the above research study. □

I agree that the chief investigator (Steve) should tell my GP about me taking part in the study, and should to tell my GP if there are any concerns about my health or welfare during the research.

□

Name of Participant _____ Signature _____ Date

_____

When complete, 1 copy for patient; 1 copy for researcher site file.

Participant Identification Number (researcher to complete): _____

Appendix 3.2 H Consent form for caregivers for people with HD

Consent Form

For Carers for people with Huntington's Disease

Title: What **Helps Me Manage My Huntington's Disease?**

Name of Researcher: Steve Smith

Please tick box to confirm

I confirm that I have read and understand the information sheet for the above study.
□

I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily.
□

I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected.

□

I understand that the sessions will be recorded, and after the researchers analyse the statements with the aid of the recording, the tape will be destroyed.
□

I agree to take part in the above research study.
□

I agree that the chief investigator (Steve) should tell my GP about me taking part in the study, and should to tell my GP if there are any concerns about my health or welfare during the research.
□

Name of Participant _____

Date _____

Signature _____

When complete, 1 copy for patient; 1 copy for researcher site file.

Patient Identification Number (Researcher to complete): _____

Appendix 3.2 I Consent form for caregivers for Health Professionals

Consent Form

For professionals caring for or supporting people with Huntington's Disease

Title: **What Helps Me Manage My Huntington's Disease?**

Name of chief investigator: Steve Smith

Please tick to confirm

I confirm that I have read and understand the information sheet dated regarding the above study.

☐

I have had the opportunity to consider the information, ask questions and am satisfied that any questions I have are answered satisfactorily.

☐

I understand that the sessions will be recorded, and after the researchers analyse the statements with the aid of the recording, the tape will be destroyed.

☐

I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason.

☐

I agree to take part in the above research study.

☐

Name of Participant _____ Profession _____

Signature _____ Date _____

When complete, 1 copy for participant; 1 copy to chief investigator in pre-paid envelope.

Participant Identification Number for this study (Researcher to complete):

_____

Appendix 3.2 J Letter to G.P.

FACULTY OF HEALTH
Edith Cavell Building
University of East Anglia
Norwich
Norfolk NR4 7TJ
Telephone
01603 597022

Dear Dr.

Re : Your Patient..... ... Participation in a research study: 'What Helps Me Manage My Huntington's Disease?

I am a lecturer in nursing at the University of East Anglia School of Nursing and Midwifery, within the Faculty of Health. I am also a PhD student and am currently undertaking the above study.

Your patient, named above was recruited via the database at the Cambridge Centre for Brain Repair at Addenbrooke's Hospital, Cambridge and has given consent to take part and for me to contact you. The patient is either a participant who has HD or is a carer for a person with HD.

I enclose an information sheet about the study, so that you know what is involved. I have contacted you now, because I recognise that speaking about Huntington's disease may have the potential to cause participants some distress. If I was to discover that your patient is very distressed I would want to contact you and inform you, and perhaps consult you for advice.

I do have many years experience of nursing people with HD, and liaising with family carers and do not feel that the risk of psychological harm will be great, but I hope you will agree that this would be a sensible precaution.

If you have any comments to make regarding this project and your patient's involvement, I would be most grateful to hear from you.

Yours sincerely

Steve Smith

Appendix 3.2 K Information for consultees

Consultee

Information sheet.

We would be very grateful if you would consider agreeing to be a consultee for **[----------Name----------]** who has Huntington's disease (HD), and is invited to take part in the study named: '***What helps me manage my Huntington's disease?'***

Why does the person need to have a consultee?

Unfortunately this person is not able to give informed consent to take part in the research. But with help s/he may be able to understand what it is about and what choosing to join in would involve. The person may be able to indicate being willing to take part.

To find out what helps people manage their Huntington's disease (HD) it is important that people severely affected by the condition are able to give their views if they want to.

A consultee is needed to give advice and offer opinion, to help the chief investigator decide about whether it is suitable to allow the person severely affected by HD to take part in the study.

What do I have to do?

If you agree to be a consultee, you should first read and make sure you understand the 'information sheet for people with HD', enclosed.

A consultee must be prepared to be consulted about the possible involvement of the potential participant in this research study who has HD. You should read and make

sure you understand the information sheet provided for this person so that you are clear about what he or she would be expected to do if he or she does take part.

If the person you are consultee for indicates being willing to take part, you may be consulted for your opinion on whether he or she would be content to take part, or whether doing so might be upsetting for him or her.

You should base this opinion on what you think the person's past and present wishes and feelings would have been about taking part in the research. For example, you could consider whether the person has previously expressed specific or general support for research about managing with HD.

You should bear in mind that you are not being asked for your personal view on taking part, but for what you think the person with HD would want to express. You could also give advice about times of the day that might be unsuitable for the person to take part. For example, the person might find this sort of activity too tiring, perhaps in the evening.

You could advise on any other factors that should be considered such as the kind of environment that might be a problem for the person, or would be more comfortable. You may advise the chief investigator (Steve) at any time if you feel that taking part is becoming too tiring for the person or for some other reason you feel they should stop being involved.

You may find it helpful to have independent advice about whether to accept this role of consultee. You could ask your GP for this advice, or a consultant with specialist knowledge of HD, or a family advisor with the Huntington's Disease Association

(HDA). Your local family advisor can be accessed via the HDA at: Tel

.....

E mail ..... HDA advisor's name

.....

If you do decide to accept the role of consultee, please sign and date the 'consultee reply slip' (enclosed) and hand it to the chief investigator (Steve Smith).

Many thanks for reading this consultee information sheet.

Appendix 3.2 L Consultee reply slip

Consultee
Reply Slip

Study title: **What helps me manage my Huntington's disease?**

Please tick box if the statement is true:

I have read and understood the Consultee information sheet

□

I understand the role of a consultee with regard to this study and am happy to accept the role

□

I have read and understood the information sheet about this research project, for people with HD.

□

I understand the research process and what the person I am consultee for will experience and be expected to do as a participant.

□

Name of consultee .....

Address.....

.....

..... Name of potential participant lacking capacity to give informed consent:

.....

Consultee- please sign to confirm the following statement is true:

……..

I have read and understand the information sheet for **people with HD**, for the study named ‘What helps me manage my Huntington’s disease?’ and I agree to accept the role of consultee for the person with HD who lacks capacity to give consent.

Signature of consultee ..... Date.....

Appendix 3.2 M Consultee confirmation document

Consultee Confirmation:

Participant's expression of willingness to take part in the study titled:

'What helps me manage my Huntington's disease?'

Please tick box if statement is true:

1) I have read the information sheet about the above study, for people with HD to the person named below, and, as best as I can, I have explained what the study involves.

□

2) The person named below has indicated to me some understanding of what the research is about and what taking part will involve

□

3) The person named below has indicated being willing to take part, and I am happy to sign to confirm that this is the case.

□

4) The person named below understands that the sessions will be audio-recorded.

□

5) The person named below agrees to the chief investigator informing their GP in the event of concerns arising about his or her health or wellbeing.

□

6) In my view it is appropriate that the person named below should be allowed to take part in the research.

□

Name of Consultee .....

Name of person with HD who lacks capacity to give informed consent :

.....

I confirm that the six numbered statements above are all true.

Signature of consultee ..... Date.....

Signature or mark of participant ..... Date .....

Appendix 3.2 N Invitation letter to people with advanced HD following approved amendment by Research Ethics Committee

Dr Sophie Duport
Royal Hospital for Neuro-Disability
Research Department
Royal Hospital for Neuro-disability
West Hill
Putney
London

SW15 3SW

Date ----------

Name Address of Recipient Here

Dear ----------

Would you like to help with some research? It is about Huntington's disease.

If so please read the other papers that tell you about it.

You may like your carer or your GP to help you understand it.

You could ask them to help you decide if it's a good idea to join in the research.

Next week, the researcher, Steve Smith, will ring to see if you are interested to take part.

If you are he will arrange to come and see you to discuss it.

If you don't want to take part, just tell Steve. It's not a problem.

Steve can be contacted by writing to: Mr Steve Smith, Lecturer

University of East Anglia, Faculty of Health, School of Nursing and Midwifery,

Edith Cavell Building, Norwich NR4 7TJ.

OR, ring and leave a message on 01603 597022, or e mail steve.smith@uea.ac.uk .

Steve will get back to you.

Very many thanks,

Yours Sincerely

Dr Sophie Duport.

Appendix 3.2 O Invitation letter to people with advanced HD following approved amendment by Research Ethics Committee

From: Sue Hill
Family Care Advisor,
Huntington's Disease Association. 01553 648 438.
sue_hill@hda.org.uk

Dear

You are invited to take part in a research project. It is to do with your role as a carer for someone who has Huntington's disease (HD), who has also been invited to take part. The person with HD was asked to give this 'carer pack' to the person they wish to be regarded as their carer for this study.

In about one week after you receive this letter, the Chief Investigator, Steve Smith, will ring you to ask if you are interested in taking part. If you are he will arrange to visit you to discuss what the study involves. Meanwhile, if you are interested, please read the enclosed information. There is no need to fill in or sign any forms yet.

If you wish to ask any questions about the project meanwhile, please contact: **Mr Steve Smith, Lecturer,**

University of East Anglia, Faculty of Health,

School of Nursing and Midwifery, Edith Cavell Building, Norwich NR4 7TJ. Or ring and leave a message on 01603 597022, or e mail steve.smith@uea.ac.uk . Steve will get back to you.

Very many thanks,

Yours Sincerely

Sue Hill,

Family Care Advisor, Huntington's Disease Association.

Appendix 3.2 P Invitation letter to people with advanced HD following approved amendment by Research Ethics Committee

From: Sue Hill

Family Care Advisor,
Huntington's Disease Association. 01553 648 438.
sue_hill@hda.org.uk

Dear

You are invited to join in with some research about Huntington's disease (HD). If you would like to know what this research is about please read the documents that are with this letter. You may want help with reading them. You could ask a carer to help, or talk to your doctor (GP). You could ask them to help you decide if it's a good idea to join in with the research.

Also, do you have a 'main' carer? I mean, someone who helps you manage your HD, who would be the right person to talk about caring for you, in this study? If so, please would you give that person the enclosed 'Carer Pack'? Thank you.

In about a week, Steve Smith, the Chief Investigator for the study, will ring you to ask if you are interested in taking part. If you are, he will arrange to visit you at your home, to discuss what is involved. Just tell Steve if you don't want to take part, it is not a problem.

If you have any questions about the study, please contact **Mr Steve Smith, Lecturer University of East Anglia, Faculty of Health, School of Nursing and Midwifery, Edith Cavell Building, Norwich NR4 7TJ.** Or ring and leave a message on 01603 597022, or e mail steve.smith@uea.ac.uk . Steve will get back to you.

Very many thanks,

Yours Sincerely

Sue Hill,

Huntington's Disease Association

Regional Care Advisor

Appendix 3.2 Q

Information Sheet For Carers of People With HD.

1. Title: **What helps me manage my Huntington's disease?**

2. **Invitation**

Would you like to join in with some research about Huntington's disease, or 'HD'? It would mean talking about your experience of caring for a person who has HD. Nothing would be done to you. A researcher called Steve who is a nurse would be asking you some questions.

You do not have to do this. Before you decide if you want to join in, it is important for you to understand what the research is for and what you would be doing.

Please take time to read the writing below, or get someone to help you or read it to you.

It is a good idea to talk to others about it if you're not sure whether to join in. You could speak to your GP (doctor) about it. Do get in touch with us if you want to know more about it.

Thank you for reading this.

3. **What is the research for?**

We hope to find out what helps people with Huntington's disease (HD) manage their HD. We want to know what people with HD and their carers and doctors and nurses and others who care think about this.

We would like to know if doctors and nurses and other professionals know what people with HD and their carers think is helpful. And we want to know how important these things are to you.

We think this will help people who care to be more helpful to people with HD in future.

4. **Why was I chosen?**

You were chosen by the person you care for who has HD. He or she was sent an invitation and a pack for carers, and was asked to give the 'carer pack' to the person they feel can best act as their main carer for this study. The person you care for with HD is one of 13 people with HD either chosen from a list kept at the Brain Repair Centre Clinic in Cambridge, or a resident at the Royal Hospital for Neuro-disability, or selected by the Huntington's Disease Association Regional Care Advisor. 10 health professionals, were also chosen.

5. **Do I have to join in?**

It is up to you to decide if you want to join in. If you decide to join in you can change your mind at any time. It will not affect the care you receive. You don't have to give a reason if you stop joining in the research.

6. **What do I have to do?**

In about a week after the person you care for, who has HD **receives an invitation**, the Chief Investigator (Steve) will ring to see if he or she, and if you are interested to join in. If you don't want to, just tell Steve. It's not a problem. If you are both interested, Steve will arrange to visit you to discuss the study, and make sure you understand what you would have to do.

If you agree to join in, Steve will ask you to sign the consent form and fill in the reply slip. You can keep a copy of each of these documents so that you can read them at any time.

If you take part, this will either mean you coming to two meetings, with other carers, or Steve visiting you twice to speak to you individually, depending on your preference. If you decide to come to the meetings, Steve will discuss with you how you will get there.

If you choose to come to the meetings**,** The first meeting will last two hours, with a break in the middle for refreshments. This meeting will be with about 9 other people who care for someone who has HD. Steve and an assistant will be there to lead the meeting and help if you're not sure what to do.

We will ask you all as a group, to agree on statements about things that you think help people to manage their HD.

The second meeting will also last two hours, with a break for refreshments. This time, people with HD, and health professionals will all be there as well as carers. This meeting will be about deciding which statements about what helps people to manage their HD are most important, and which are not so important.

On the day we will be there to help to make sure you understand how to do it. But there will be no 'right' or 'wrong' answers. We want your ideas.

If you choose instead for Steve to visit you individually, you will be asked to make statements about what is helpful to people with HD on the first visit, and to decide which statements you feel are most important on the second visit.

There is one more thing we would like you to help with. After the meetings are finished Steve and the research team will work to make a report of the results of the study. We would like to send you a copy of a draft of this report, for you to comment on. You could point out anything you do or don't agree with. Then a final report will be written, and you will be given a copy. The Huntington's Disease Association will also get a copy.

An important thing to note is that we will audio record the meetings. This is to help us remember what was said as we write about the study. It will help us if we are not sure what any written statements mean. After the report is written the tape will be destroyed. H

7. **Are there any risks to me?**

Talking about how caring for someone who has HD has affected you could upset you and make you think about some things you like to forget.

If you told us something that made us think the person you care for is not being treated well, or is at risk of harm, we would tell the person's GP. The GP would then help us decide if we need to do anything else, to protect the person. If you told us something that made us think either you or the person you care for is thinking about self harm or suicide, we would tell the GP, who again, would help us decide if we need to do more to protect either or both of you.

8. **Could joining in the study do me some good?**

On the other hand, it might feel good to talk about things that help the person you care for to manage. And what helps you. You might feel good that this will help

people in future, to know what helps people with HD. But, the research will not help you personally.

9. **Will my taking part in this study be kept confidential?**

Steve will keep the forms you sign with your name and other details locked safely. He may publish some of your ideas but will not tell anyone your personal details. Steve will tape record what you say to remind him later. The tape will be kept locked safely and will be destroyed when the research is finished.

If you decide to join in group discussions, the others in the room will be asked to keep what you say confidential. But we cannot guarantee they will. You do not have to say anything that you don't want others to know.

10. **What will happen to the results of the research study?**

We will try to publish results in research journals to do with health. Also, we will give the Huntington's Disease Association a report, to print in the newsletter if they wish.

The chief investigator (Steve) will talk about the study at meetings about HD.

11. **Who is organising and funding the research?**

This study is part of a PhD project for the chief investigator (Steve), and is paid for by the University of East Anglia, School of Nursing Midwifery.

12. **Who has reviewed the study?**

The study has been reviewed by experienced health researchers at the University of East Anglia. It has been approved by the Local Research Ethics Committee. It will be monitored by a research team including experienced researchers, a person with

HD, and a carer, and a Family Care Advisor from the Huntington's Disease Association.

13. **Contact for Further Information**

Very many thanks for taking part in this study if you do decide to. If you wish to discuss it further or ask any questions about it, please contact the chief investigator:

Steve Smith
University of East Anglia,
Faculty of Health,
School of Nursing and Midwifery
Edith Cavell Building
Norwich NR4 7TJ.
Tel: 01603 59 7022 (leave a message if not there and Steve will get back to you) Or e mail: steve.smith@uea.ac.uk

What should I do if I'm not happy with any aspect of the study?

You could contact the Huntington's Disease Association and to talk to the local Family Advisor, Sue Hill, tel: 01353 648438, or e mail: sue_hill@hda.org.uk to discuss any concerns, or you could make a complaint to Rachel Mold, Faculty Research Manager, tel: 01603 5973949, e mail: r.mold@uea.ac.uk.

Declaration:

I have read and understood this information sheet.

Signed _____ Name (Print)

_____

Date _____

Please Note: You should sign and return one copy of this information sheet and a consent form, and keep a 2nd copy of each.

Reply Slip

If you have decided that you wish to take part in the study: 'What Helps Me Manage My Huntington's Disease?', please complete this slip and send it to Steve Smith with the consent form, signed and dated, and the information sheet, also signed and dated (one copy only of each document).

Full Name _____ Date of Birth _____

Gender Male Female (please delete as applicable).

Address_____

_____

How long have you cared for the person with HD taking part in this study?

_____

If you have cared for others with HD, how many years have you spent caring for a person with HD in total? _____

What is your relationship to the person with HD that you are caring for?

_____

Name Address and phone number of your GP.

_____

Phone number or e mail address for the researcher (Steve) to contact you on, if needed, to discuss arrangements for the meetings:

_____

Signed _____ Date _____

Appendix 3.2 R

Information Sheet For People With HD.

1. Title: **What helps me manage my Huntington's disease?**

2. **Invitation**

Would you like to join in with some research about Huntington's disease, or 'HD'? It would mean talking about your experience of caring for a person who has HD. Nothing would be done to you. A researcher called Steve who is a nurse would be asking you some questions.

You do not have to do this. Before you decide if you want to join in, it is important for you to understand what the research is for and what you would be doing.

Please take time to read the writing below, or get someone to help you or read it to you.

It is a good idea to talk to others about it if you're not sure whether to join in. You could speak to your GP (doctor) about it. Do get in touch with us if you want to know more about it.

Thank you for reading this.

3. **What is the research for?**

We hope to find out what helps people with Huntington's disease (HD) manage their HD. We want to know what people with HD and their carers and doctors and nurses and others who care think about this.

We would like to know if doctors and nurses and other professionals know what people with HD and their carers think is helpful. And we want to know how important these things are to you.

We think this will help people who care to be more helpful to people with HD in future.

4. **Why was I chosen?**

You are one of 13 people with HD either chosen from a list kept at the Brain Repair Centre Clinic in Cambridge, or are a resident at the Royal Hospital for Neuro-disability, or were selected by the Huntington's Disease Association Regional Care Advisor. 10 health professionals, were also chosen.

5. **Do I have to join in?**

It is up to you to decide if you want to join in. If you decide to join in you can change your mind at any time. It will not affect the care you receive. You don't have to give a reason if you stop joining in the research.

6. **What do I have to do?**

In about a week after you receive an invitation, the Chief Investigator (Steve) will ring to see if you are interested to join in. If you don't want to, just tell Steve. It's not a problem. If you are interested, Steve will arrange to visit you to discuss the study, and make sure you understand what you would have to do.

If you agree to join in, Steve will ask you to sign the consent form and fill in the reply slip. You can keep a copy of each of these documents so that you can read them at any time.

If you take part, this will either mean you coming to two meetings, with other people with HD, or Steve visiting you twice to speak to you individually, depending on your preference. If you decide to come to the meetings, Steve will discuss with you how you will get there.

If you choose to come to the meetings**,** The first meeting will last two hours, with a break in the middle for refreshments. This meeting will be with about 9 other people who care for someone who has HD. Steve and an assistant will be there to lead the meeting and help if you're not sure what to do.

We will ask you all as a group, to agree on statements about things that you think help people to manage their HD.

The second meeting will also last two hours, with a break for refreshments. This time, people with HD, and health professionals will all be there as well as carers. This meeting will be about deciding which statements about what helps people to manage their HD are most important, and which are not so important.

On the day we will be there to help to make sure you understand how to do it. But there will be no 'right' or 'wrong' answers. We want your ideas.

If you choose instead for Steve to visit you individually, you will be asked to make statements about what is helpful to people with HD on the first visit, and to decide which statements you feel are most important on the second visit.

There is one more thing we would like you to help with. After the meetings are finished Steve and the research team will work to make a report of the results of the study. We would like to send you a copy of a draft of this report, for you to comment on. You could point out anything you do or don't agree with. Then a final report will

be written, and you will be given a copy. The Huntington's Disease Association will also get a copy.

An important thing to note is that we will audio record the meetings. This is to help us remember what was said as we write about the study. It will help us if we are not sure what any written statements mean. After the report is written the tape will be destroyed. H

7. **Are there any risks to me?**

Talking about how HD has affected you could upset you and make you think about some things you like to forget.

If you told us something that made us think the person you care for is not being treated well, or is at risk of harm, we would tell the person's GP. The GP would then help us decide if we need to do anything else, to protect the person. If you told us something that made us think either you or the person you care for is thinking about self harm or suicide, we would tell the GP, who again, would help us decide if we need to do more to protect either or both of you.

8. **Could joining in the study do me some good?**

On the other hand, it might feel good to talk about things that help you to manage. You might feel good that this will help people in future, to know what helps people live with HD.

But, the research will not help you personally.

9. **Will my taking part in this study be kept confidential?**

Steve will keep the forms you sign with your name and other details locked safely. He may publish some of your ideas but will not tell anyone your personal details. Steve will tape record what you say to remind him later. The tape will be kept locked safely and will be destroyed when the research is finished.

If you decide to join in group discussions, the others in the room will be asked to keep what you say confidential. But we cannot guarantee they will. You do not have to say anything that you don't want others to know.

10. **What will happen to the results of the research study?**

We will try to publish results in research journals to do with health. Also, we will give the Huntington's Disease Association a report, to print in the newsletter if they wish.

The chief investigator (Steve) will talk about the study at meetings about HD.

11. **Who is organising and funding the research?**

This study is part of a PhD project for the chief investigator (Steve), and is paid for by the University of East Anglia, School of Nursing Midwifery.

12. **Who has reviewed the study?**

The study has been reviewed by experienced health researchers at the University of East Anglia. It has been approved by the Local Research Ethics Committee. It will be monitored by a research team including experienced researchers, a person with HD, and a carer, and a Family Care Advisor from the Huntington's Disease Association.

13. **Contact for Further Information**

Very many thanks for taking part in this study if you do decide to. If you wish to discuss it further or ask any questions about it, please contact the chief investigator:

Steve Smith
University of East Anglia,
Faculty of Health,
School of Nursing and Midwifery
Edith Cavell Building
Norwich NR4 7TJ.
Tel: 01603 59 7022 (leave a message if not there and Steve will get back to you) Or e mail: steve.smith@uea.ac.uk

What should I do if I'm not happy with any aspect of the study?

You could contact the Huntington's Disease Association and to talk to the local Family Advisor, Sue Hill, tel: 01353 648438, or e mail: sue_hill@hda.org.uk to discuss any concerns, or you could make a complaint to Rachel Mold, Faculty Research Manager, tel: 01603 5973949, e mail: r.mold@uea.ac.uk.

Declaration:

I have read and understood this information sheet.

Signed _____ Name (Print) _____

Date _____

Please Note: You should sign and return one copy of this information sheet and a consent form, and keep a 2nd copy of each.

Reply Slip

If you have decided that you wish to take part in the study: 'What Helps Me Manage My Huntington's Disease?', please complete this slip and send it to Steve Smith with the consent form, signed and dated, and the information sheet, also signed and dated (one copy only of each document).

Full Name _____ Date of Birth _____

Gender Male Female (please delete as applicable).

Address_____

_____

How long have you been affected by symptoms of HD? _____

Name Address and phone number of your GP.

_____

Phone number or e mail address for the researcher (Steve) to contact you on, if needed, to discuss arrangements for the meetings:

_____

Signed _____ Date _____

Appendix 3.3 Data Entry Forms

Entry form prioritising task

ID number participant:

DATE ----------

Please enter the number of the statements as they have been prioritized.

	least important 1	**2**	**3**	**4**	**most important 5**
statement 1					
2					
3					
4					
5					
6					
7					
8					
9					
10					
11					
12					
13					

14					
15					
(min.) 16					
17					
18					

Appendix 3.3 Data Entry Form (clustering task, Page 1)

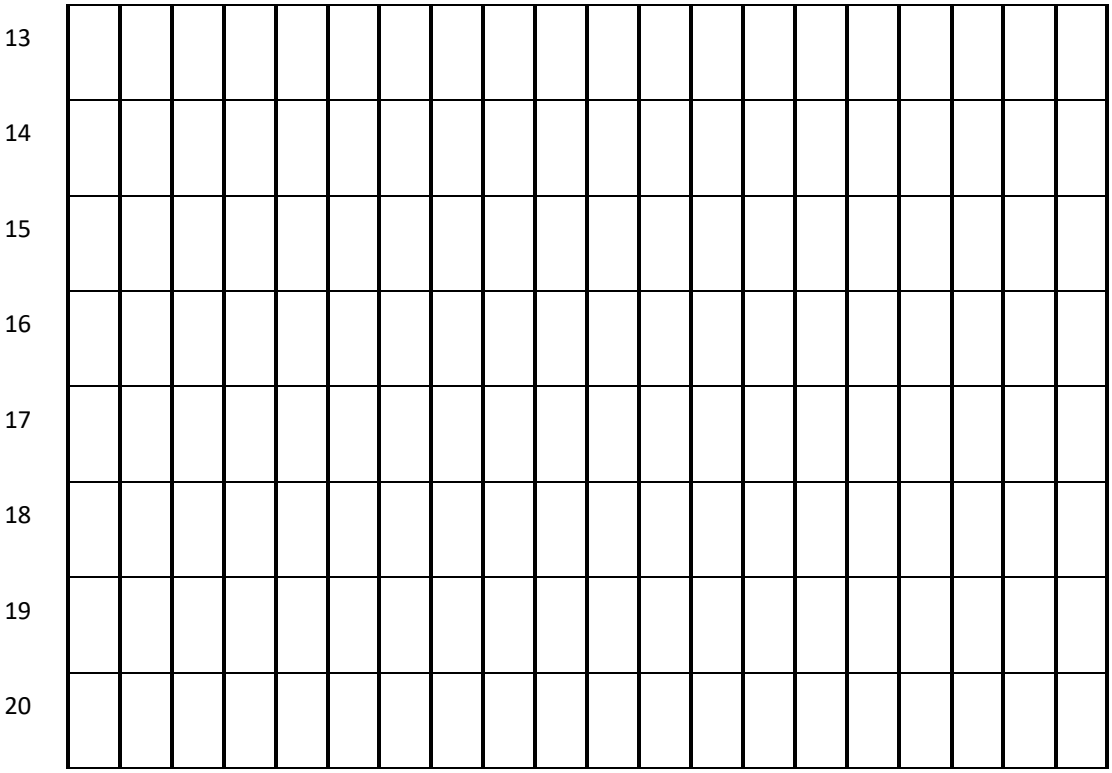
ID number participant:

Name:

Date:

Please enter the number of the statements as they have been clustered.

	cluster 1	**cluster 2**	**cluster 3**	**cluster 4**	**cluster 5**	**cluster 6**	**cluster 7**	**cluster 8**	**cluster 9**	**cluster 10**	**cluster 11**	**cluster 12**	**cluster 13**	**cluster 14**	**cluster 15**	**cluster 16**	**cluster 17**	**cluster 18**	**cluster 19**	**cluster 20**
statement 1																				
2																				
3																				
4																				
(min) 5																				
6																				
7																				
8																				
9																				
10																				
11																				
12																				


13
14
15
16
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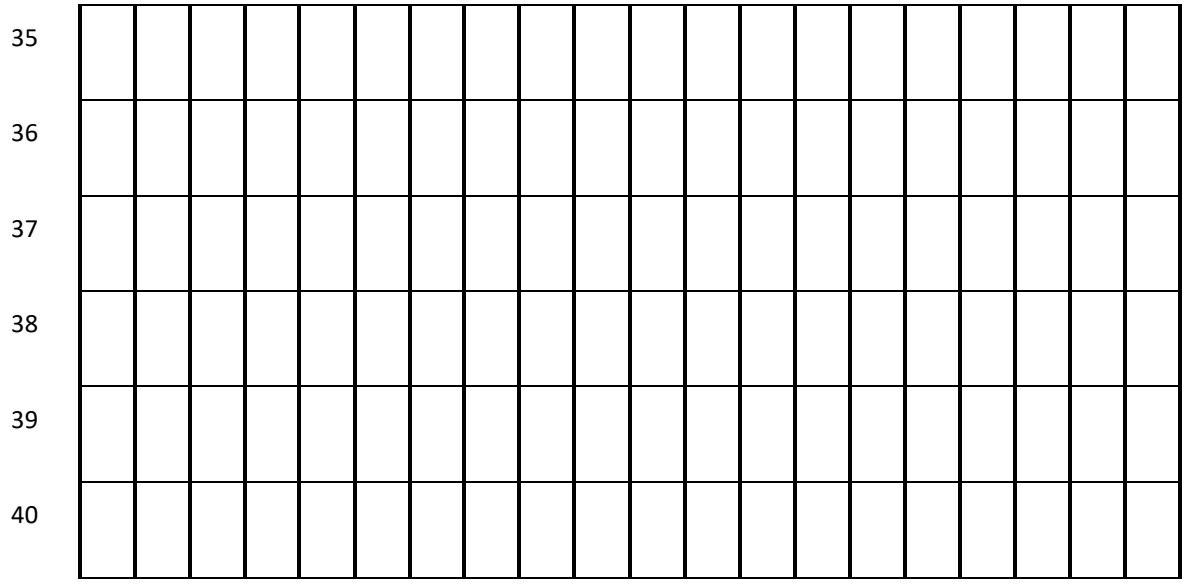
19
20

Appendix 3.3 Entry form clustering task, page 2

ID number participant:

DATE ----------

	cluster 1	cluster 2	cluster 3	cluster 4	cluster 5	cluster 6	cluster 7	cluster 8	cluster 9	cluster 10	cluster 11	cluster 12	cluster 13	cluster 14	cluster 15	cluster 16	cluster 17	cluster 18	cluster 19	cluster 20
statement 21																				
22																				
23																				
24																				
25																				
26																				
27																				
28																				
29																				
30																				
31																				
32																				
33																				
34																				


35
36
37
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39
40

Appendix 3.4 Test for Normal Distribution

Tests of Normality

	Kolmogorov-Smirnov[a]			Shapiro-Wilk		
	Statistic	df	Sig.	Statistic	df	Sig.
Having medications to help sleep	.162	64	.000	.880	64	.000
Medication against chorea	.184	64	.000	.868	64	.000
Having appropriately timed medication reviews	.187	64	.000	.916	64	.000
Medication against chorea to help me with activities of living	.235	64	.000	.849	64	.000
Knowing that the effects of medications for HD are available	.224	64	.000	.883	64	.000
Knowing side effectsof drugs for HD can be serious	.236	64	.000	.870	64	.000
Recognition that antipsychotic medication can be helpful in HD	.165	64	.000	.898	64	.000
Knowing that sedative medication will not be given solely to manage behaviour	.175	64	.000	.882	64	.000
Having good quality healthcare professionals and carers	.211	64	.000	.855	64	.000
Having community understanding	.218	64	.000	.891	64	.000

a. Lilliefors Significance Correction

Appendix 4.1 Table depicting each of the numbered 94 statements, showing which statements each data point on the map, Fig 4.1, represents.

1	Having medicines to help sleep
2	Medication against chorea
3	Having appropriately timed reviews of medication
4	Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.
5	knowing that the effects of medications for HD are variable
6	Knowing that side effects of drugs for HD can be serious
7	recognition that antipsychotic medication can be helpful in HD
8	Knowing that sedative medication will not be given solely to manage behaviour
9	Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation
10	Having community understanding
11	Regular and timely access to health and social care assessment
12	Having a positive outlook on life
13	Living for today
14	Making choices about care including end of life care in advance and having the choice respected
15	Assessment of management of aggression if it occurs.
16	If obsessive behaviour and impulsivity are recognised as serious symptoms of HD
17	If there is expert assessment and treatment of mental health problems (eg. Depression, anxiety) occurring along with HD
18	If professionals help the person with HD to get a life
19	Having help to socialise
20	If the person with HD has help to be able to get out and about
21	Knowing in advance the detailed plans for what will happen today and / or at forthcoming events
22	Timeliness in access to care homes
23	Not being cared for in an inappropriate setting (eg. A psychiatric or elderly setting where staff don't know about HD).
24	Having a familiar daily routine.
25	Being prudent with money
26	Spending money while he or she is able to
27	Having someone to sort out his or her access to financial benefits
28	Direct access to cash benefits to spend on what is needed now
29	If they are enabled to maintain the roles that they feel suit their gender
30	If employers make reasonable adjustments to enable continued employment
31	If they accept the diagnosis.
32	Having accessible town, transport and shop facilities
33	Having alcohol
34	Having cigarettes
35	Being involved in a Huntington's Disease Association support group
36	Having the opportunity to be involved in research
37	Having a stable relationship with a partner
38	If informal carers have training in HD
39	Having access to the right level of information about HD at the right time
40	Having support from the Huntington's Disease Association
41	Access to professionals who are expert in HD

42	Access to a multi-disciplinary team that is expert in HD
43	Being referred to a specialist National HD centre
44	Effective multi-disciplinary working
45	Effective interagency working
46	When service provision is planned for when it will be needed, rather than thought about once things have already got difficult
47	If professionals follow up patients who do not keep appointments
48	Having the goal of a normal life when considering the use of medication
49	The availability of a genetic test
50	Pre-test and post-test counselling
51	Not knowing whether other family members are genetically positive for HD
52	Knowing if other family members are genetically positive for HD
53	Availability of pre-implantation diagnosis
54	Timely introduction of tube feeding
55	Having assistance to eat and drink
56	Having family and friends who offer practical support
57	If there is availability of food that is appetising and manageable
58	Having practical strategies to enable him or her to eat out without embarrassment
59	Practical advice on specific foods and textures
60	If he or she is able to talk freely and openly about HD within the family
1	Having a supportive network of family and friends
62	Having appropriate training to use equipment
63	Having a smoking aid
64	Having aids to enable daily living (eg. Aids that help with Communication, mobility, furniture, bathroom and falls prevention).
65	Having timely access to appropriate aids and adaptations
66	Regular timely assessment for aids and adaptations
67	A prompt diagnosis
68	If professionals work with all members of the family
69	If people with HD know the role of each professional
70	Speech and language therapy involvement to help with swallowing problems
71	If visiting carers are flexible regarding the specific type of care they provide in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).
72	If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).
73	Participating in intellectual activities
74	Participating in physical activities
75	Being able to take up hobbies and pastimes
76	Still being able to drive.
77	Being able to undertake domestic activities such as cooking, gardening and cleaning
78	Being able to go away from the care home to engage in community activities
79	Having meaningful relationships with other care home residents
80	If a care home environment is made to feel as home should feel
81	Continuity of health and social care professionals
82	Having continuity of care staff, (eg, professional carers in homes or that visit at

	home).
83	Having opportunities for meaningful conversation
84	Recognition that facial and neck pain can be a problem in HD.
85	Speech and language therapy involvement in maintaining ability to communicate.
86	Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).
87	Having a choice about seeing a male or a female GP
88	If care is available where the person with HD needs it.
89	care is available when the person with HD needs it.
90	If carers are listened to as an adult and believed and respected.
91	If they are listened to as an adult and believed and respected
92	If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.
93	If the family carer has a break from looking after him or her.
94	If there is information about HD for health professionals in general medical settings

Table presenting 94 statements in order of priority according to mean scores, for all participants.

Rank	Mean rating	Statement name
1	4.12	Access to professionals who are expert in HD
2	4.09	Having a supportive network of family and friends
3	3.95	Having a familiar daily routine.
4	3.92	Speech and language therapy involvement to help with swallowing problems
5	3.92	If care is available when the person with HD needs it.
6	3.88	Having family and friends who offer practical support
7	3.86	If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.
8	3.82	Having a stable relationship with a partner
9	3.82	Access to a multi-disciplinary team that is expert in HD
10	3.80	If the family carer has a break from looking after him or her.
11	3.80	Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation
12	3.75	Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.
13	3.75	If care is available where the person with HD needs it.
14	3.72	If there is expert assessment and treatment of mental health problems (eg. Depression, anxiety) occurring along with HD
15	3.72	Not being cared for in an inappropriate setting (eg. A psychiatric or elderly setting where staff don't know about HD).
16	3.72	If they are listened to as an adult and believed and respected.
17	3.63	Having aids to enable daily living (eg. Aids that help with communication, mobility, furniture, bathroom and falls prevention).
18	3.62	If the person with HD has help to be able to get out and about
19	3.62	Speech and language therapy involvement in maintaining ability to communicate
20	3.59	Making choices about care including end of life care in advance and having the choice respected
21	3.58	Having support from the Huntington's Disease Association.
22	3.58	Having a positive outlook on life
23	3.57	If he or she is able to talk freely and openly about HD within the family
24	3.52	Being referred to a specialist National HD centre
25	3.52	Having continuity of care staff, (eg, professional carers in homes or that visit at home).
26	3.49	Having opportunities for meaningful conversation
27	3.49	If carers are listened to as an adult and believed and respected
28	3.48	Medication against chorea
29	3.48	Being involved in a Huntington's Disease Association support group
30	3.46	Regular and timely access to health and social care assessment
31	3.43	When service provision is planned for when it will be needed, rather than thought about once things have already got difficult
32	3.35	Having help to socialise
33	3.35	Having assistance to eat and drink
34	3.32	A prompt diagnosis

35	3.31	Continuity of health and social care professionals
36	3.28	Having timely access to appropriate aids and Adaptations
37	3.26	If they accept the diagnosis.
38	3.22	Having appropriately timed reviews of medication
39	3.22	Having medicines to help sleep
40	3.22	Having access to the right level of information about HD at the right time
41	3.18	Regular timely assessment for aids and adaptations
42	3.17	If informal carers have training in HD
43	3.17	If there is availability of food that is appetising and Manageable
44	3.13	Having the goal of a normal life when considering the use of medication
45	3.11	Pre-test and post-test counselling
46	3.11	If professionals work with all members of the family
47	3.11	If there is information about HD for health professionals in general medical settings
48	3.09	Effective multi-disciplinary working
49	3.08	Living for today
50	3.06	Assessment of management of aggression if it occurs.
51	3.05	Participating in physical activities
52	3.03	Having someone to sort out his or her access to financial benefits
53	3.03	The availability of a genetic test
54	3.02	If obsessive behaviour and impulsivity are recognised as serious symptoms of HD
55	3.02	If a care home environment is made to feel as home should feel
56	2.98	If visiting carers are flexible regarding the specific type of care they provide
57	2.91	If professionals help the person with HD to get a life
58	2.85	recognition that antipsychotic medication can be helpful in HD
59	2.83	Knowing in advance the detailed plans for what will happen today and / or at forthcoming events
60	2.83	Having the opportunity to be involved in research
61	2.83	Effective interagency working
62	2.83	Being able to take up hobbies and pastimes
63	2.83	Practical advice on specific foods and textures
64	2.78	Having practical strategies to enable him or her to eat out without embarrassment
65	2.78	Being able to go away from the care home to engage in community activities
66	2.77	Having community understanding
67	2.71	Having accessible town, transport and shop facilities
68	2.71	If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).
69	2.66	If employers make reasonable adjustments to enable continued employment
70	2.63	Direct access to cash benefits to spend on what is needed now
71	2.63	If professionals follow up patients who do not keep Appointments
72	2.63	If people with HD know the role of each professional
73	2.62	Availability of pre-implantation diagnosis
74	2.60	Participating in intellectual activities
75	2.57	Access to experimental treatments (not as part of research, but to find a

		treatment that works for the person with HD).
76	2.55	Timely introduction of tube feeding
77	2.55	Being able to undertake domestic activities such as cooking, gardening and cleaning
78	2.54	Timeliness in access to care homes
79	2.51	Knowing that sedative medication will not be given solely to manage behaviour
80	2.51	If they are enabled to maintain the roles that they feel suit their gender
81	2.45	Knowing that side effects of drugs for HD can be Serious
82	2.42	knowing that the effects of medications for HD are variable
83	2.35	Having appropriate training to use equipment
84	2.28	Still being able to drive.
85	2.23	Knowing if other family members are genetically positive for HD
86	2.13	Recognition that facial and neck pain can be a problem in HD.
87	2.12	Spending money while he or she is able to
88	2.00	Having meaningful relationships with other care home Residents
89	1.89	Having alcohol
90	1.86	Not knowing whether other family members are genetically positive for HD
91	1.85	Having a choice about seeing a male or a female GP
92	1.83	Having cigarettes
93	1.68	Being prudent with money
94	1.66	Having a smoking aid

Appendix 4.3 Table presenting 94 statements in order of priority according to mean scores, for people with HD

Rank	Rating	Statement
1	4.32	Access to professionals who are expert in HD
2	4.11	Having a positive outlook on life
3	4.11	Having a supportive network of family and friends
4	4.05	If care is available where the person with HD needs it.
5	4.00	Having a stable relationship with a partner
6	3.95	Speech and language therapy involvement to help with swallowing problems
7	3.89	If the family carer has a break from looking after him or her.
8	3.84	Having family and friends who offer practical support
9	3.79	If there is expert assessment and treatment of mental health problems (eg. Depression, anxiety) occurring along with HD
10	3.79	If they are listened to as an adult and believed and respected.
11	3.74	Speech and language therapy involvement in maintaining ability to communicate
12	3.68	Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.
13	3.68	Having support from the Huntington's Disease Association.
14	3.67	Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation
15	3.63	Having help to socialise
16	3.63	If carers are listened to as an adult and believed and Respected
17	3.58	Making choices about care including end of life care in advance and having the choice respected
18	3.58	Not being cared for in an inappropriate setting (eg. A psychiatric or elderly setting where staff don't know about HD).
19	3.58	Access to a multi-disciplinary team that is expert in HD
20	3.58	The availability of a genetic test
21	3.58	Having continuity of care staff, (eg, professional carers in homes or that visit at home).
22	3.58	If care is available when the person with HD needs it.
23	3.58	If there is information about HD for health professionals in general medical settings
24	3.53	Medication against chorea
25	3.53	Continuity of health and social care professionals
26	3.47	Having a familiar daily routine.
27	3.47	If they accept the diagnosis.
28	3.47	If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.
29	3.42	If the person with HD has help to be able to get out and about
30	3.42	Still being able to drive.
31	3.32	If obsessive behaviour and impulsivity are recognised as serious symptoms of HD
32	3.32	Being involved in a Huntington's Disease Association support group
33	3.32	When service provision is planned for when it will be needed, rather than thought about once things have already got difficult
34	3.32	If there is availability of food that is appetising and Manageable
35	3.28	If visiting carers are flexible regarding the specific type of care they

		provide
36	3.26	Participating in physical activities
37	3.26	Being able to take up hobbies and pastimes
38	3.26	Having opportunities for meaningful conversation
39	3.21	Having someone to sort out his or her access to financial benefits
40	3.21	Having the opportunity to be involved in research
41	3.21	Being referred to a specialist National HD centre
42	3.21	A prompt diagnosis
43	3.17	Having the goal of a normal life when considering the use of medication
44	3.16	Regular and timely access to health and social care assessment
45	3.16	Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).
46	3.11	Living for today
47	3.11	Having aids to enable daily living (eg. Aids that help with Communication, mobility, furniture, bathroom and falls prevention).
48	3.05	recognition that antipsychotic medication can be helpful in HD
49	3.05	Effective multi-disciplinary working
50	3.05	If professionals work with all members of the family
51	3.00	If informal carers have training in HD
52	3.00	Participating in intellectual activities
53	2.95	If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).
54	2.89	Having medicines to help sleep
55	2.89	Having assistance to eat and drink
56	2.89	Practical advice on specific foods and textures
57	2.89	If he or she is able to talk freely and openly about HD within the family
58	2.89	Being able to undertake domestic activities such as cooking, gardening and cleaning
59	2.84	Having access to the right level of information about HD at the right time
60	2.84	Pre-test and post-test counselling
61	2.84	Having practical strategies to enable him or her to eat out without embarrassment
62	2.79	Having appropriately timed reviews of medication
63	2.79	Assessment of management of aggression if it occurs.
64	2.79	If a care home environment is made to feel as home should feel
65	2.74	knowing that the effects of medications for HD are variable
66	2.74	Having community understanding
67	2.74	Having accessible town, transport and shop facilities
68	2.68	Being able to go away from the care home to engage in community activities
69	2.63	If people with HD know the role of each professional
70	2.58	Knowing that sedative medication will not be given solely to manage behaviour
71	2.58	Spending money while he or she is able to
72	2.58	Direct access to cash benefits to spend on what is needed now
73	2.58	Knowing if other family members are genetically positive for HD
74	2.53	If professionals follow up patients who do not keep appointments
75	2.53	Availability of pre-implantation diagnosis
76	2.47	If employers make reasonable adjustments to enable continued

		employment
77	2.47	Regular timely assessment for aids and adaptations
78	2.42	Knowing that side effects of drugs for HD can be serious
79	2.42	Effective interagency working
80	2.37	If they are enabled to maintain the roles that they feel suit their gender
81	2.37	Having timely access to appropriate aids and Adaptations
82	2.37	Recognition that facial and neck pain can be a problem in HD.
83	2.32	Knowing in advance the detailed plans for what will happen today and / or at forthcoming events
84	2.32	Timeliness in access to care homes
85	2.32	Having alcohol
86	2.26	Timely introduction of tube feeding
87	2.26	Having appropriate training to use equipment
88	2.21	If professionals help the person with HD to get a life
89	2.00	Not knowing whether other family members are genetically positive for HD
90	2.00	Having a choice about seeing a male or a female GP
91	1.95	Being prudent with money
92	1.89	Having cigarettes
93	1.89	Having meaningful relationships with other care home residents
94	1.63	Having a smoking aid

Appendix 4 4. Table presenting 94 statements in order of priority according to mean scores, for caregivers

Rank	Mean rating	Statement
1	4.23	If care is available when the person with HD needs it.
1	4.19	Having a familiar daily routine.
3	4.19	If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.
4	4.15	Having a supportive network of family and friends
5	4.12	Being referred to a specialist National HD centre
6	4.04	If the person with HD has help to be able to get out and about
7	4.04	Having a stable relationship with a partner
8	4.04	Access to professionals who are expert in HD
9	4.04	Having family and friends who offer practical support
10	4.00	Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation
11	3.88	Having support from the Huntington's Disease Association.
12	3.88	Speech and language therapy involvement to help with swallowing problems
13	3.88	Having continuity of care staff, (eg, professional carers in homes or that visit at home).
14	3.88	If the family carer has a break from looking after him or her.
15	3.85	Being involved in a Huntington's Disease Association support group
16	3.81	If he or she is able to talk freely and openly about HD within the family
17	3.77	Access to a multi-disciplinary team that is expert in HD
18	3.73	Having aids to enable daily living (eg. Aids that help with Communication, mobility, furniture, bathroom and falls prevention).
19	3.69	Not being cared for in an inappropriate setting (eg. A psychiatric or elderly setting where staff don't know about HD).
20	3.62	Having a positive outlook on life
21	3.62	Having access to the right level of information about HD at the right time
22	3.62	If care is available where the person with HD needs it.
23	3.58	If there is expert assessment and treatment of mental health problems (eg. Depression, anxiety) occurring along with HD
24	3.58	Having timely access to appropriate aids and adaptations
25	3.58	If carers are listened to as an adult and believed and respected
26	3.54	Speech and language therapy involvement in maintaining ability to communicate
27	3.50	Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.
28	3.46	Having help to socialise
29	3.46	Having opportunities for meaningful conversation
30	3.38	If informal carers have training in HD
31	3.38	Having assistance to eat and drink
32	3.38	Regular timely assessment for aids and adaptations
33	3.38	A prompt diagnosis
34	3.35	Continuity of health and social care professionals
35	3.31	Participating in physical activities
36	3.27	Regular and timely access to health and social care assessment

37	3.27	If they accept the diagnosis.
38	3.23	Having medicines to help sleep
39	3.23	If professionals help the person with HD to get a life
40	3.23	When service provision is planned for when it will be needed, rather than thought about once things have already got difficult
41	3.20	Having appropriately timed reviews of medication
42	3.19	Having someone to sort out his or her access to financial benefits
43	3.19	If they are listened to as an adult and believed and respected.
44	3.16	Making choices about care including end of life care in advance and having the choice respected
45	3.15	Assessment of management of aggression if it occurs.
46	3.15	If a care home environment is made to feel as home should feel
47	3.12	Medication against chorea
48	3.12	If there is availability of food that is appetising and Manageable
49	3.08	If professionals work with all members of the family
50	3.08	If visiting carers are flexible regarding the specific type of care they provide
51	3.08	If there is information about HD for health professionals in general medical settings
52	3.04	If obsessive behaviour and impulsivity are recognised as serious symptoms of HD
53	3.00	Living for today
54	3.00	Having the opportunity to be involved in research
55	3.00	Having the goal of a normal life when considering the use of medication
56	3.00	Pre-test and post-test counselling
57	2.92	The availability of a genetic test
58	2.92	Being able to take up hobbies and pastimes
59	2.77	Knowing in advance the detailed plans for what will happen today and / or at forthcoming events
60	2.77	Having accessible town, transport and shop facilities
61	2.77	Effective interagency working
62	2.77	If people with HD know the role of each professional
63	2.73	Direct access to cash benefits to spend on what is needed now
64	2.73	If professionals follow up patients who do not keep appointments
65	2.73	Having appropriate training to use equipment
66	2.72	Practical advice on specific foods and textures (n=25)
67	2.69	Having community understanding
68	2.65	Effective multi-disciplinary working
69	2.65	Being able to go away from the care home to engage in community activities
70	2.65	Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).
71	2.58	Availability of pre-implantation diagnosis
72	2.54	Knowing that side effects of drugs for HD can be Serious
73	2.54	Having practical strategies to enable him or her to eat out without embarrassment
74	2.54	If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).
75	2.50	Timeliness in access to care homes

76	2.50	Participating in intellectual activities
77	2.46	Timely introduction of tube feeding
78	2.42	recognition that antipsychotic medication can be helpful in HD
79	2.35	If they are enabled to maintain the roles that they feel suit their gender
80	2.31	Knowing that sedative medication will not be given solely to manage behaviour
81	2.31	If employers make reasonable adjustments to enable continued employment
82	2.27	knowing that the effects of medications for HD are variable
83	2.27	Being able to undertake domestic activities such as cooking, gardening and cleaning
84	2.15	Having meaningful relationships with other care home residents
85	2.00	Spending money while he or she is able to
86	2.00	Knowing if other family members are genetically positive for HD
87	2.00	Recognition that facial and neck pain can be a problem in HD.
88	1.92	Not knowing whether other family members are genetically positive for HD
89	1.88	Having alcohol
90	1.77	Still being able to drive.
91	1.58	Having cigarettes
92	1.50	Being prudent with money
93	1.50	Having a choice about seeing a male or a female GP
94	1.46	Having a smoking aid

Appendix 4.5 Table presenting 94 statements in order of priority according to mean scores, for Health Care Professionals

Rank	Mean rating	Statement
1	4.35	If they are listened to as an adult and believed and respected.
2	4.15	Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.
3	4.15	Making choices about care including end of life care in advance and having the choice respected
4	4.10	Having a familiar daily routine.
5	4.10	Access to a multi-disciplinary team that is expert in HD
6	4.05	Access to professionals who are expert in HD
7	4.00	Regular and timely access to health and social care assessment
8	4.00	Having a supportive network of family and friends
9	4.00	Having aids to enable daily living (eg. Aids that help with Communication, mobility, furniture, bathroom and falls prevention).
10	3.95	Speech and language therapy involvement to help with swallowing problems
11	3.90	Medication against chorea
12	3.90	Not being cared for in an inappropriate setting (eg. A psychiatric or elderly setting where staff don't know about HD).
13	3.90	If he or she is able to talk freely and openly about HD within the family
14	3.85	If there is expert assessment and treatment of mental health problems (eg. Depression, anxiety) occurring along with HD
15	3.85	If care is available when the person with HD needs it.
16	3.80	When service provision is planned for when it will be needed, rather than thought about once things have already got difficult
17	3.80	If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.
18	3.75	Having assistance to eat and drink
19	3.75	Having timely access to appropriate aids and adaptations
20	3.75	Having opportunities for meaningful conversation
21	3.70	Effective multi-disciplinary working
22	3.70	Having family and friends who offer practical support
23	3.65	Having appropriately timed reviews of medication
24	3.65	Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation
25	3.65	If care is available where the person with HD needs it.
26	3.60	Regular timely assessment for aids and adaptations
27	3.60	Speech and language therapy involvement in maintaining ability to communicate
28	3.60	If the family carer has a break from looking after him or her.
29	3.50	Having medicines to help sleep
30	3.50	Pre-test and post-test counselling
31	3.40	Knowing in advance the detailed plans for what will happen today and / or at forthcoming events
32	3.35	Having a stable relationship with a partner
33	3.35	A prompt diagnosis
34	3.30	If employers make reasonable adjustments to enable continued

		employment
35	3.30	Effective interagency working
36	3.25	If the person with HD has help to be able to get out and about
37	3.25	Having the goal of a normal life when considering the use of medication
38	3.25	If carers are listened to as an adult and believed and respected
39	3.20	recognition that antipsychotic medication can be helpful in HD
40	3.20	Assessment of management of aggression if it occurs.
41	3.20	If professionals work with all members of the family
42	3.15	Living for today
43	3.15	If professionals help the person with HD to get a life
44	3.15	Being involved in a Huntington's Disease Association support group
45	3.10	Having support from the Huntington's Disease Association.
46	3.10	If there is availability of food that is appetising and manageable
47	3.05	If they accept the diagnosis.
48	3.05	If informal carers have training in HD
49	3.05	Having access to the right level of information about HD at the right time
50	3.05	Being referred to a specialist National HD centre
51	3.05	Having practical strategies to enable him or her to eat out without embarrassment
52	3.05	Being able to go away from the care home to engage in community activities
53	3.05	If a care home environment is made to feel as home should feel
54	3.05	Continuity of health and social care professionals
55	3.00	Having a positive outlook on life
56	3.00	Having continuity of care staff, (eg, professional carers in homes or that visit at home).
57	2.95	Having help to socialise
58	2.95	Timely introduction of tube feeding
59	2.90	Having community understanding
60	2.90	Practical advice on specific foods and textures
61	2.85	If they are enabled to maintain the roles that they feel suit their gender
62	2.80	Timeliness in access to care homes
63	2.75	Availability of pre-implantation diagnosis
64	2.70	Knowing that sedative medication will not be given solely to manage behaviour
65	2.70	If obsessive behaviour and impulsivity are recognised as serious symptoms of HD
66	2.70	If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).
67	2.70	If there is information about HD for health professionals in general medical settings
68	2.65	Having someone to sort out his or her access to financial benefits
69	2.65	The availability of a genetic test
70	2.60	Having accessible town, transport and shop facilities
71	2.60	If professionals follow up patients who do not keep appointments
72	2.60	If visiting carers are flexible regarding the specific type of care they provide
73	2.60	Being able to undertake domestic activities such as cooking, gardening and cleaning

74	2.55	Direct access to cash benefits to spend on what is needed now
75	2.50	Participating in physical activities
76	2.45	If people with HD know the role of each professional
77	2.35	Knowing that side effects of drugs for HD can be serious
78	2.35	Participating in intellectual activities
79	2.30	knowing that the effects of medications for HD are variable
80	2.30	Being able to take up hobbies and pastimes
81	2.25	Having the opportunity to be involved in research
82	2.20	Knowing if other family members are genetically positive for HD
83	2.15	Having a choice about seeing a male or a female GP
84	2.10	Having cigarettes
85	2.05	Recognition that facial and neck pain can be a problem in HD.
86	1.95	Having appropriate training to use equipment
87	1.95	Having a smoking aid
88	1.90	Having meaningful relationships with other care home residents
89	1.90	Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).
90	1.85	Spending money while he or she is able to
91	1.85	Still being able to drive.
92	1.65	Being prudent with money
93	1.65	Not knowing whether other family members are genetically positive for HD
94	1.50	Having alcohol

Appendix 4.6 Table presenting 94 statements in order of priority according to mean scores, for the Nursing Home Subgroup (Caregivers and Health Professionals only - no people with HD included) (n=19, 11 professionals, 8 caregivers).

Rank	Mean Rating	Statement
1	4.32	Access to a multi-disciplinary team that is expert in HD
2	4.26	Having a familiar daily routine.
3	4.21	Access to professionals who are expert in HD
4	4.11	Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.
5	4.05	Having a supportive network of family and friends
6	3.95	Speech and language therapy involvement to help with swallowing problems
7	3.89	Medication against chorea
8	3.89	If he or she is able to talk freely and openly about HD within the family
9	3.89	Having aids to enable daily living (eg. Aids that help with Communication, mobility, furniture, bathroom and falls prevention).
10	3.84	If care is available where the person with HD needs it.
11	3.84	If care is available when the person with HD needs it.
12	3.84	If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.
13	3.79	Having assistance to eat and drink
14	3.74	Being referred to a specialist National HD centre
15	3.68	If they accept the diagnosis.
16	3.68	Having a stable relationship with a partner
17	3.63	If the person with HD has help to be able to get out and about
18	3.63	Not being cared for in an inappropriate setting (eg. A psychiatric or elderly setting where staff don't know about HD).
19	3.63	If they are listened to as an adult and believed and respected.
20	3.58	Being involved in a Huntington's Disease Association support group
21	3.58	When service provision is planned for when it will be needed, rather than thought about once things have already got difficult
22	3.58	Having family and friends who offer practical support
23	3.58	Speech and language therapy involvement in maintaining ability to communicate
24	3.58	If the family carer has a break from looking after him or her.
25	3.53	Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation
26	3.53	If carers are listened to as an adult and believed and respected
27	3.42	Having medicines to help sleep
28	3.42	recognition that antipsychotic medication can be helpful in HD
29	3.42	Regular and timely access to health and social care assessment
30	3.42	Having access to the right level of information about HD at the right time
31	3.42	Having timely access to appropriate aids and adaptations
32	3.42	Having continuity of care staff, (eg, professional carers in homes or that visit at home).

33	3.37	Regular timely assessment for aids and adaptations
34	3.37	Having opportunities for meaningful conversation
35	3.32	If there is expert assessment and treatment of mental health problems (eg. Depression, anxiety) occurring along with HD
36	3.32	Having support from the Huntington's Disease association.
37	3.32	Having the goal of a normal life when considering the use of medication
38	3.26	Having appropriately timed reviews of medication
39	3.26	If professionals help the person with HD to get a life
40	3.21	Having help to socialise
41	3.21	Knowing in advance the detailed plans for what will happen today and / or at forthcoming events
42	3.16	Assessment of management of aggression if it occurs.
43	3.16	Timely introduction of tube feeding
44	3.16	If there is availability of food that is appetising and Manageable
45	3.16	Participating in physical activities
46	3.16	If a care home environment is made to feel as home should feel
47	3.11	Making choices about care including end of life care in advance and having the choice respected
48	3.05	If employers make reasonable adjustments to enable continued employment
49	3.05	A prompt diagnosis
50	3.05	Being able to go away from the care home to engage in community activities
51	3.00	Effective multi-disciplinary working
52	3.00	Pre-test and post-test counselling
53	3.00	Practical advice on specific foods and textures
54	3.00	Continuity of health and social care professionals
55	2.94	Having a positive outlook on life
56	2.89	Living for today
57	2.89	Availability of pre-implantation diagnosis
58	2.84	Having accessible town, transport and shop facilities
59	2.84	If informal carers have training in HD
60	2.84	Effective interagency working
61	2.84	If professionals work with all members of the family
62	2.84	If visiting carers are flexible regarding the specific type of care they provide
63	2.74	If obsessive behaviour and impulsivity are recognised as serious symptoms of HD
64	2.74	Having practical strategies to enable him or her to eat out without embarrassment
65	2.68	Having someone to sort out his or her access to financial benefits
66	2.63	Knowing that sedative medication will not be given solely to manage behaviour
67	2.63	Having community understanding
68	2.63	If they are enabled to maintain the roles that they feel suit their gender
69	2.63	If professionals follow up patients who do not keep appointments
70	2.63	Being able to take up hobbies and pastimes
71	2.63	If there is information about HD for health professionals in general medical settings
72	2.58	If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can

		help the person with HD, if the person with HD doesn't want help from the professional carer).
73	2.53	Direct access to cash benefits to spend on what is needed now
74	2.47	Timeliness in access to care homes
75	2.42	Having the opportunity to be involved in research
76	2.42	If people with HD know the role of each professional
77	2.37	Being able to undertake domestic activities such as cooking, gardening and cleaning
78	2.32	knowing that the effects of medications for HD are variable
79	2.32	Knowing if other family members are genetically positive for HD
80	2.32	Participating in intellectual activities
81	2.32	Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).
82	2.26	The availability of a genetic test
83	2.26	Recognition that facial and neck pain can be a problem in HD.
84	2.21	Knowing that side effects of drugs for HD can be Serious
85	2.21	Having a choice about seeing a male or a female GP
86	2.11	Having cigarettes
87	2.11	Having appropriate training to use equipment
88	2.11	Still being able to drive.
89	2.11	Having meaningful relationships with other care home Residents
90	1.95	Not knowing whether other family members are genetically positive for HD
91	1.84	Spending money while he or she is able to
92	1.84	Having a smoking aid
93	1.79	Being prudent with money
94	1.68	Having alcohol

Appendix 4.7 Table presenting 94 statements in order of priority according to mean scores, for the Community-Based Subgroup (Caregivers and Health Professionals only - no people with HD included) (n=27: 18 caregivers, 9 professionals).

Rank	Mean Rating	Statement
1	4.23	If care is available when the person with HD needs it.
2	4.15	If family carers are believed when they report reaching a tipping point, where care at home is no longer manageable.
3	4.12	Having family and friends who offer practical support
4	4.12	Having a familiar daily routine.
5	4.08	Having a supportive network of family and friends
6	4.04	Having good quality healthcare professionals and carers who are good communicators; share information; stay calm and avoid confrontation
7	4.00	Making choices about care including end of life care in advance and having the choice respected
8	3.96	If there is expert assessment and treatment of mental health problems (eg. Depression, anxiety) occurring along with HD
9	3.96	Speech and language therapy involvement to help with swallowing problems
10	3.96	Not being cared for in an inappropriate setting (eg. A psychiatric or elderly setting where staff don't know about HD).
11	3.88	If the family carer has a break from looking after him or her.
12	3.88	If he or she is able to talk freely and openly about HD within the family
13	3.88	Access to professionals who are expert in HD
14	3.88	Having a stable relationship with a partner
15	3.85	Having timely access to appropriate aids and adaptations
16	3.81	If they are listened to as an adult and believed and respected.
17	3.77	Having aids to enable daily living (eg. Aids that help with Communication, mobility, furniture, bathroom and falls prevention).
18	3.77	Regular and timely access to health and social care assessment
19	3.73	Access to a multi-disciplinary team that is expert in HD
20	3.69	Having opportunities for meaningful conversation
21	3.69	Having support from the Huntington's Disease Association.
22	3.69	If the person with HD has help to be able to get out and about
23	3.62	Being referred to a specialist National HD centre
24	3.62	A prompt diagnosis
25	3.58	Regular timely assessment for aids and adaptations
26	3.58	Having continuity of care staff, (eg, professional carers in homes or that visit at home).
27	3.58	Having a positive outlook on life
28	3.54	Speech and language therapy involvement in maintaining ability to communicate
29	3.54	If informal carers have training in HD
30	3.50	Medication against chorea to help the person with HD do activities such as sleep, have a sex life, reduce distress, reduce fatigue and carry out activities of living.
31	3.50	Being involved in a Huntington's Disease Association support group
32	3.48	Having appropriately timed reviews of medication
33	3.46	If care is available where the person with HD needs it.

34	3.46	When service provision is planned for when it will be needed, rather than thought about once things have already got difficult
35	3.42	If carers are listened to as an adult and believed and respected
36	3.42	If professionals work with all members of the family
37	3.38	Having access to the right level of information about HD at the right time
38	3.38	Continuity of health and social care professionals
39	3.38	Pre-test and post-test counselling
40	3.31	Having assistance to eat and drink
41	3.27	Effective multi-disciplinary working
42	3.23	Having medicines to help sleep
43	3.19	Having help to socialise
44	3.19	Effective interagency working
45	3.19	Assessment of management of aggression if it occurs.
46	3.19	The availability of a genetic test
47	3.12	If there is availability of food that is appetising and manageable
48	3.12	If professionals help the person with HD to get a life
49	3.12	Having someone to sort out his or her access to financial Benefits
50	3.12	If a care home environment is made to feel as home should feel
51	3.12	Living for today
52	3.08	Medication against chorea
53	3.08	If there is information about HD for health professionals in general medical settings
54	3.00	Having the goal of a normal life when considering the use of Medication
55	3.00	If obsessive behaviour and impulsivity are recognised as serious symptoms of HD
56	2.96	Knowing in advance the detailed plans for what will happen today and / or at forthcoming events
57	2.88	If they accept the diagnosis.
58	2.85	If visiting carers are flexible regarding the specific type of care they provide
59	2.85	If people with HD know the role of each professional
60	2.81	Timeliness in access to care homes
61	2.81	Having community understanding
62	2.77	Having practical strategies to enable him or her to eat out without embarrassment
63	2.77	If professionals follow up patients who do not keep appointments
64	2.77	Having the opportunity to be involved in research
65	2.73	Participating in physical activities
66	2.72	Practical advice on specific foods and textures
67	2.65	Direct access to cash benefits to spend on what is needed now
68	2.65	Having accessible town, transport and shop facilities
69	2.65	Knowing that side effects of drugs for HD can be serious
70	2.62	Being able to go away from the care home to engage in community activities
71	2.62	If professional carers are flexible about who they help in the household (for example, being prepared to help children, so that the spouse can help the person with HD, if the person with HD doesn't want help from the professional carer).
72	2.58	Having appropriate training to use equipment
73	2.58	If they are enabled to maintain the roles that they feel suit their gender
74	2.58	If employers make reasonable adjustments to enable Continued

		employment
75	2.58	Being able to take up hobbies and pastimes
76	2.50	Being able to undertake domestic activities such as cooking, gardening and cleaning
77	2.42	Participating in intellectual activities
78	2.42	Availability of pre-implantation diagnosis
79	2.38	Timely introduction of tube feeding
80	2.38	Knowing that sedative medication will not be given solely to manage behaviour
81	2.31	recognition that antipsychotic medication can be helpful in HD
82	2.27	knowing that the effects of medications for HD are variable
83	2.23	Access to experimental treatments (not as part of research, but to find a treatment that works for the person with HD).
84	2.00	Having meaningful relationships with other care home residents
85	1.96	Knowing if other family members are genetically positive for HD
86	1.92	Spending money while he or she is able to
87	1.80	Recognition that facial and neck pain can be a problem in HD.
88	1.73	Not knowing whether other family members are genetically positive for HD
89	1.73	Having alcohol
90	1.62	Still being able to drive.
91	1.62	Having cigarettes
92	1.58	Having a smoking aid
93	1.46	Having a choice about seeing a male or a female GP
94	1.38	Being prudent with money