Experiences of Caring for a Partner Diagnosed with Huntington’s Disease: An Interpretative Phenomenological Analysis

Thesis submitted in part fulfilment of the degree of

Doctorate in Clinical Psychology

(DClinPsy)

University of Leicester

By

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Declaration

I can confirm that this thesis and the research reported within it is my original work. It was written and submitted in part fulfilment of the degree of Doctorate in Clinical Psychology (DClinPsy). It has not been submitted for any other degree or academic qualification.
Experiences of Caring for a Spouse with Huntington’s Disease Through Pre and Post Clinical Diagnosis: An Interpretative Phenomenological Analysis

Sarah Crozier

Abstract

Huntington’s disease (HD) is a neurodegenerative genetic condition for which a predictive genetic test by mutation analysis has been available since 1993. However, whilst revealing the future presence of the disease, testing may have an adverse psychological impact given that the disease is progressive, incurable and ultimately fatal.

The current literature review aimed to explore the published evidence base examining the psychological impact of predictive genetic testing for HD. Based on the synthesis of eight research studies, the process of predictive genetic testing was not found to be psychological neutral with fluctuating levels of distress irrelevant of test result. Methodological weaknesses were identified highlighting the needs of individuals not accessing testing or follow-up services, warranting further assessment.

The research study sought to understand the lived experiences of partner carers of individuals diagnosed with HD throughout the disease trajectory. Qualitative interviews were conducted with six carers whose partners were currently accessing HD services.

Analysis using Interpretative Phenomenological Analysis (IPA) revealed five super-ordinate themes with 18 sub-themes allowing for idiosyncrasies of respondents’ experiences to be accounted for. Themes were considered in relation to previous literature within HD and parallel fields with clinical implications highlighted. A need for further exploratory and subsequent quantitative research of phenomena was warranted and recommended.

The critical appraisal provides the Researcher’s reflective account of the research process.
Acknowledgements

I would like to thank the six respondents who willingly volunteered their time to take part in this research and share their stories and experiences. Without their honest and thoughtful contributions this research would not have been possible.

I would also like to thank my research supervisor, Dr Noelle Robertson at the University of Leicester, for her support and guidance throughout the last three years. I am particularly thankful to my field supervisor and clinical psychologist\(^1\) within the Huntington’s disease service of which this research was based. Your passion and interest in working with Huntington’s disease and the families affected by it was truly inspiring. To the Huntington’s disease service community nurses I would like to show my appreciation for your help with recruitment of participants.

Finally I would like to thank my family and friends, particularly my husband and mum. You have never failed to support me in my journey to fulfilling my dreams and goals. Thank you for your continuous encouragement and unconditional love, alongside my much needed distractions and reminders to look after myself.

Thank you all so much.

\(^1\) To ensure anonymity of the service and research respondents, it is with regret that this supervisor could not be named
## Word Counts

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Transcript 2: ‘Ruth’
Transcript 3: ‘Brendan’
Transcript 4: ‘Peter’
Transcript 5: ‘Glenda’
Transcript 6: ‘Henry’

Names given are pseudonyms to maintain participant anonymity

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3 Names given are pseudonyms to maintain participant anonymity
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Part 1

Literature Review

The Psychological Impact of Predictive Genetic Testing for Huntington's Disease: A Systematic Review of the Literature

This literature review has been prepared in line with the Journal of Genetic Counseling guidelines.

Guidelines for authors can be found in Appendix A.
Abstract

Huntington’s disease (HD) is a neurodegenerative genetic condition for which a predictive genetic test by mutation analysis has been available since 1993. This has permitted identification of individuals who are asymptomatic genetic carriers of the disease. Yet the use of genetic tests, whilst revealing the future presence of the disease, may have an adverse psychological impact given that the disease is progressive, incurable and ultimately fatal.

This review seeks to systematically explore the psychological impact of genetic testing for individuals undergoing pre-symptomatic mutation analysis. Three databases (Medline, PsycInfo and Scopus) were interrogated for studies utilising standardised measures to assess psychological impact following predictive genetic testing for HD. From 100 papers initially identified, eight articles were eligible for inclusion.

Psychological impact of predictive genetic testing was not found to be associated with test result. No detrimental effect of predictive genetic testing on non-carriers was found, although the process was not found to be psychologically neutral. Fluctuation in levels of distress was found over time for both carriers and non-carriers alike. Methodological weaknesses of published literature were identified, notably the needs of individuals not requesting genetic testing, as well as inadequate support for individuals registering elevated distress and declining post-test follow-up. Further assessment of these vulnerable individuals is warranted to establish the extent and type of future psychological input.

Key Words: genetic testing, predictive testing, Huntington’s disease, psychological impact, systematic review
1. Introduction

Huntington’s disease (HD) is an *autosomal dominantly inherited* neurodegenerative condition affecting one person per 10,000 in the UK alone (Evans *et al.*, 2013). The average age of onset for the disease is 40 years with a typical disease trajectory of 10-20 years until death (Myers, 2004). HD is characterised by a triad of impairments in movement, cognition and affect. Many people who develop the disease are aware of its incidence within their family and may be hypervigilant to early signs and symptoms in themselves. Prior to clinical diagnosis changes to personality, mood and cognitive abilities may be present. Affected individuals will then begin to present with chorea and difficulties with balance which will progressively decline to the latter stage of the disease where bradykinesia and dystonia are observed with frequent secondary difficulties such as heart failure and pneumonia. Currently the disease is incurable although treatments may improve quality of life.

Although HD was first thoroughly described in 1872 (Huntington, 1872), it was only in the 1980’s that genetic markers for HD were located within chromosome four (Gusella *et al.*, 1983). Through a process of tracing inheritance markers across generations of affected and unaffected family members, the first genetic testing by *linkage analysis* was developed and offered to those *at-risk* in 1986. Initially, linkage analysis was only

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4 Italicised words and phrases are explained in the glossary of terms provided in Appendix B
considered for research purposes but was later used in clinical settings offering at-risk individuals testing within 95% accuracy of likely future development of HD (Huntington’s Disease Society of America, 2001).

Linkage analysis was replaced in 1993 by direct mutation analysis following the isolation of the gene responsible for HD (Huntington’s Disease Collaborative Research Group, 1993). HD was definitively found to result from an expansion of the trinucleotide repeat (CAG) coding for a protein involved in nerve cell function. Essentially, whilst healthy individuals will have between 11 and 26 repeats of the CAG trinucleotide, those who go on to develop HD have a CAG repeat length of greater than 40. Rare outcomes of testing are results that fall within the reduced penetrance range (36-39 CAG repeats), or those with intermediate alleles (27-35 CAG repeats). Mutation analysis testing allowed assessment for the presence of the mutation gene, giving individuals up to 100% certainty of their status as a carrier of the condition (Evers-Kiebooms & Decruyenaere, 1998).

Prior to the availability of a specific genetic test, between 56% and 81% of individuals at-risk of HD expressed a desire for testing (Koller & Davenport, 1984; Tyler & Harper, 1983), however after testing was offered, figures suggested a maximum of 20% uptake (Craufurd et al., 1989; Quaid & Morris, 1993). This discrepancy may be explained by the complexity implicit in making a decision to undergo predictive genetic testing. Diverse motives may operate not least reducing uncertainty, shaping reproductive
choices, responding to reactions of family members and practical planning, as well as fears of coping in the event of a positive test result (Decruyenaere et al., 1997).

Psychological issues too are significant in shaping the decision, as well as being evidenced in reactions to an unfavourable genetic test, after which intense distress and potential for suicidal risk has been documented (Kessler, 1987; Kessler et al., 1987). Specific threats to an individual’s health encountered as a result of undergoing predictive genetic testing for HD include the potential for definitive genetic certainty of developing the disease, as well as a response to the absence of effective treatments and inevitable decline (Goody et al., 2006). For these reasons, in conditions such as HD with little hope of ameliorating interventions, the ethical and psychological implications of predictive genetic testing is of paramount importance. Such understanding permits sensitivity to those who may be psychologically at-risk and consideration of possible interventions (Salkovskis & Rimes, 1997).

Knowledge of genetic status has been revealed to have both a positive and negative impact on an individual’s life (Duncan et al., 2008). Circumscribed qualitative research exploring the experiences of individuals who have undergone predictive genetic testing for HD has revealed regret and distress associated with anticipated life change, limitations and a loss of hope, alongside increased appreciation of life and relief from uncertainty.
(Hagberg et al., 2011). Adverse psychological impact has been suggested, irrespective of test result, from either the adjustment to the inevitable progression to diagnosis for carriers, or adaptation to survivor guilt for non-carriers (Hayden & Bombard, 2005). The latter term encompasses the burden of remaining healthy whilst others will test positive and develop the disease. Psychological difficulties post-testing have also been attributed to over-optimism regarding the impact of favourable test results in as many as 10% of individuals identified as non-carriers (Huggins et al., 1992).

Various psychological models have endeavoured to encapsulate reactions to predictive genetic testing. Notable amongst these is the Common Sense Model of self-regulation of health and illness, (Leventhal et al., 1998) in both its application and development of an evidence base. This model advances the idea of parallel processes of appraisal and coping, using both cognitive and emotional strategies, as a means of reducing distress caused by a threat to health (Leventhal et al., 2001). Using illness representations (the cognitions implicit in appraisal), the threat of a disease is appraised regarding its cause, controllability and consequence over time with health behaviours argued as the means by which threat is managed. Concurrently emotional processes trigger alternative, usually unconscious, coping strategies to manage fear and uncertainty. Using this model, HD can be constructed as uncontrollable in its development and fatal in consequence, making it likely to be appraised as highly threatening and therefore highly distressing. A distressing period of “knowing about not
knowing," can lead an at-risk individual to seek out predictive genetic
testing (Konran, 2003) and this process of applying for testing can
therefore be seen as a coping strategy for managing an at-risk status by
providing control to the individuals through seeking certainty and
knowledge (Gooding et al., 2006).

An equally useful perspective on the threat to those who are genetically at-
risk is offered by Rolland and Williams (2005) who, in a family systems
model argue that appraisals of health threat encompass a three way
interaction of disease-specific typology, time phase and functioning.
Disease-specific typology is based on an individual's appraisal of disease
likelihood, clinical severity, time of onset and availability of treatments. For
HD, with its 50% inheritance pattern, clinical impact in middle-adulthood,
lack of treatment options and subsequent fatality, disease appraisals are
likely to be more intensely negative than for other genetic conditions. The
identification of a genetically at-risk population from predictive genetic
testing has prompted the definition of disease within the model to be
broadened to include the time prior to clinical diagnosis. In addition to the
disease time phases highlighted in the family systems illness model
(Rolland, 1984), non-symptomatic disease time phases of awareness,
pretesting crisis, test/post-testing crisis and long term adaption have also
been identified. The psychological impact of disease may therefore
resonate throughout these time phases; beyond predictive genetic testing
but before clinical diagnosis.
From both these models, an individual's representation of a disease, and its meaning, may influence coping strategies and functioning and has framed the exploration of psychological reactions of individuals undergoing predictive genetic testing. To date, findings have been equivocal. A systematic review, undertaken over a decade ago examining global psychological consequences of predictive genetic testing (of which HD respondents were non-separate) identified no abnormally high or increased levels of distress in individuals assessed up to three years post-test, however a sub-group of individuals were identified as having high levels of anxiety or depression post-testing (Broadstock et al., 2000). By contrast, a narrative review of psychological consequences from HD-specific predictive genetic testing highlighted differential psychological impact between individuals found to be carriers and those found to be non-carriers in the short-term (Meiser & Dunn, 2000). More recently the adverse psychological impact of predictive genetic testing has been suggested irrespective of test result, although psychological distress appears to be manifest along differing time trajectories (Almqvist et al., 2003).

### 1.1 Aims of the Current Review

Theory and a circumscribed evidence base suggest that there may be differential psychological impact of predictive genetic testing dependent on disease type. This finding, allied with research revealing significantly higher
levels of distress in individuals undergoing predictive genetic testing for HD in comparison to other genetic conditions (Dudok DeWit et al., 1998; 1997), prompted the researcher’s initial focus on the specific psychological impact of predictive genetic testing in individuals at-risk of HD.

Whilst the impact of predictive genetic testing in HD specifically has been explored in two non-systematic reviews (Meiser & Dunn, 2000; Hayden & Bombard, 2005), both are limited by a focus on predictive genetic testing conducted by the discontinued linkage analysis, albeit alongside mutation analysis. A singular focus on the psychological impact of testing via mutation analysis is warranted given some evidence of elevated depression, pre- and post-test (Adam et al., 1995; Codori et al., 1997). Explanatory theories may also imply that those receiving predictive results through mutation analysis may experience more adverse reactions given the definitive nature of the result.

Reviews to date have also reflected a lack of published empirical research, particularly examining comparisons between individuals choosing to undergo predictive testing and those who do not, and an absence of studies addressing cognitive or behavioural consequences of predictive testing (Broadstock et al., 2000). Additional limitations are evident because of the circumscribed periods of post-test follow-up that were conducted because of the relatively recent adoption of mutation analysis at the time they were undertaken. Since it is now a decade since mutation analysis
was introduced, the current review was felt timely to examine longer-term follow-up data, to enrich evaluation of the psychological impact of predictive genetic testing.

The aim of the current review was therefore to review systematically the published evidence base exploring the psychological impact of the process of pre-symptomatic predictive mutation analysis testing for HD in individuals at-risk of the disease. Examination of impacts is hoped to guide the development and delivery of clinically supportive services and shape the focus of future research.
2. Method

2.1 Search Strategy

A thorough examination of the literature available was completed with adherence to a systematic search process. An initial scoping search developed a focus with the formation of search strings. This permitted the identification of literature addressing the main aims of the current review. Search strings were grouped to address the main focus areas including: psychological impact (psycholog*; impact; effect; consequence), genetic testing (genetic; predictive; testing; screening) and HD (HD; Huntington*).

Searches, conducted in November 2013, and repeated in March 2014, used Medline, PsycInfo and Scopus databases to ensure a range of medical and psychological literature was included. Reference lists from identified papers were examined to identify further relevant literature as well as consultation with researchers in the HD field. To validate the results of the search strings, references were compared with those included within previously published narrative reviews (Meiser & Dunn, 2000; Hayden & Bombard, 2005).

Articles published prior to 1993 were excluded, consistent with the time at which the mutation analysis test was introduced. Searches were limited to peer-reviewed articles in the English language. The search strings entered
into each database and the number of articles found can be seen in Appendix C.

From the three databases, a total of 157 articles were identified, 80 from Scopus, 26 from PsycInfo and 51 from Medline. A further three articles were identified from consultation with known researchers in the field. With duplicates removed, a total of 100 abstracts remained. Titles and abstracts were screened against the eligibility criteria with 33 warranting full text appraisals.

2.2 Eligibility Criteria

The full texts of articles were coded for eligibility by the principal researcher with articles excluded if they were:

- Qualitative – requirement of a standardised outcome measure of psychological impact from genetic testing

- Reviews, meta-analyses or narrative accounts of knowledge
Other exclusion criteria included:

- Using a non HD at-risk sample or a HD at-risk sample not separable from other genetically inherited conditions
- Using a child sample
- Using confirmatory genetic, prenatal genetic or linkage analysis testing
- Case studies or conference abstracts

### 2.3 Quality Appraisal and Data Extraction

Full articles were independently appraised with regards to conformity to Strengthening the Reporting of Observational studies in Epidemiology (STROBE) combined checklist (Von Elm et al., 2008). Checklists were used to guide the researcher’s judgements to ensure quality assessment and evaluation of bias. Data extraction forms were independently used to code all articles for inclusion (Appendix D). Half the articles to be included were randomly selected and independently coded by the supervising researcher for purposes of reliability and validity. Little discrepancy was evident and was clarified through discussion to achieve consensus.
2.4 Eligible Papers

A total of eight articles were included within the current review as summarised in Appendix E. Reasons for article exclusion were: use of alternative genetic testing methods other than predictive mutation analysis testing (10); no use of standardised measures of the psychological impact of genetic testing (five); and analysis with differing genetic testing procedures or genetically inherited conditions not permitting the assessment of data pertaining solely to HD (10). A summary of the article selection process is given in figure 1.

Figure 1. PRISMA Flowchart (Moher et al., 2009) showing number of articles excluded at each stage of eligibility screening.
3. Results

3.1 Study Characteristics

The main characteristics of the studies included in this review are shown in Table I. Studies were conducted across six countries: Belgium (Decruyenaere et al., 1996); France (Gargiulo et al., 2009); USA (Horowitz et al., 2001); Sweden (Larsson et al., 2006; Wahlin et al., 2000); Germany (Licklederer et al., 2008); and the Netherlands (Timman et al., 2004; Witjes-Ané et al., 2002). Recruitment of participants in all papers was via a single centre genetic clinic associated with a university hospital, with the exception of one study in which recruitment was through contacts with multiple professionals working in the field and response to an advert placed in a HD-specific newsletter (Licklederer et al., 2008).

One study employed a retrospective design (Gargiulo et al., 2009) and a further study used a between-subjects design (Licklederer et al., 2008). All other studies used a prospective, repeated measures design with baseline measures compared with between two and five follow-up points. Study sample sizes ranged from 34 (Wahlin et al., 2000) to 134 (Witjes-Ané et al., 2002) with a mean age ranging from 36.9 (Wahlin et al., 2000) to 41.9 years (Gargiulo et al., 2009). The percentage of female participants within the studies ranged from 50% (Wahlin et al., 2000) to 68% (Horowitz et al., 2001).
Table I. Characteristics of studies assessing psychological impact of genetic testing in HD

<table>
<thead>
<tr>
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<tr>
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<td>Non-Carrier</td>
<td>Carrier</td>
<td></td>
</tr>
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<td>31</td>
<td>22</td>
<td>Baseline, 1m, 1yr</td>
</tr>
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<td>Gargiulo et al., 2009 (France)</td>
<td>62</td>
<td>57</td>
<td>3m – 9yr</td>
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<tr>
<td>Horowitz et al., 2001 (USA)</td>
<td>44</td>
<td>Asymptomatic</td>
<td>Symptom-atic</td>
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<tr>
<td>Larsson et al., 2006 (Sweden)</td>
<td>35</td>
<td>58</td>
<td>Baseline, 2m, 6m, 12m, 24m</td>
</tr>
<tr>
<td>Licklederer et al., 2008 (Germany)</td>
<td>52</td>
<td>Asymptomatic</td>
<td>Symptom-atic</td>
</tr>
<tr>
<td>Timman et al., 2004 (Netherlands)</td>
<td></td>
<td></td>
<td>61</td>
</tr>
<tr>
<td>Wahlin et al., 2000 (Sweden)</td>
<td>21</td>
<td>13</td>
<td>Baseline, 2m, 6m, 12m, 24m</td>
</tr>
<tr>
<td>Witjes-Ané et al., 2002 (Netherlands)</td>
<td>88</td>
<td>46</td>
<td>Baseline (0-61m), 18m</td>
</tr>
</tbody>
</table>

wk: weeks; m: months; yr: years
BDI: Beck Depression Inventory; STAI: State Trait Anxiety Inventory; BHS: Beck Hopelessness Scale;
MMPI: Minnesota Multiphasic Personality Inventory; IES: Impact of Events Scale;
SIBS: Self Injurious Behaviour Scale; GHQ-30: General Health Questionnaire-30; GSI: Global Severity Index;
SF-12: Short Form-12; UHDRS: Unified Huntington's Disease Rating Scale

Constructs used to assess the psychological impact of genetic testing included: depression and hopelessness (seven studies), anxiety (two studies), distress (three studies), psychological well-being (five studies) and self-injurious/suicide tendency (two studies). Depression and hopelessness were measured using the Beck Depression Inventory (BDI: Beck et al., 1961) or Beck Hopelessness Scale (BHS: Beck, 1974). Anxiety was measured by the State Trait Anxiety Inventory (STAI: Spielberger,
1983) with measures of general and specific, state and trait anxiety. Distress was measured with the Impact of Events Scale (IES: Horowitz et al., 1979) giving cognitive and affective indices associated with intrusion and avoidance. Psychological well-being was determined through use of Baron’s Ego Strength Scale from the Minnesota Multiphasic Personality Inventory (MMPI: Graham, 1987), General Health Questionnaire-30 (GHQ-30: Goldberg & Williams, 1988), Global Severity Index (GSI) of the Brief Symptom Inventory (BSI: Derogatis & Melisaratos, 1983) or the Short Form Health Questionnaire (SF-12: Bullinger & Kirchberger, 1998). The Self Injurious Behaviour Scale (SIBS: Fox et al., 1989) was used to measure self-injurious or suicide tendency. The Unified Huntington’s Disease Rating Scale (UHDRS: Huntington Study Group, 1996) was used in one study as measure of affect and behaviour change looking at depression, low self-esteem, anxiety, suicidal thoughts, obsessions, compulsions, irritable behaviour, disruptive or aggressive behaviour, delusions and hallucinations.

3.2 Psychological Impact of HD Genetic Test Result

Eight studies were identified which compared the psychological impact of HD genetic testing contingent on test result as summarised in Table II. Three studies with a follow-up of up to ten years post-test have highlighted no significant difference between individuals given a carrier status and
those given a *non-carrier* status (Decruyenaere et al., 1996; Timman et al., 2004; Wahlin et al., 2000).

No study suggested a detrimental psychological effect of genetic testing for HD when the test outcome was favourable (revealing non-carrier status) compared to when test outcome was undesirable (revealing carrier status). Some studies reported a positive psychological impact in non-carriers, with a significantly lower level of hopelessness and distress (Gargiulo et al., 2009), depression and low self-esteem, alongside non-significant trends for aggression and compulsive behaviours up to nine years post-test (Witjes-Ané et al., 2002). However, neither study accounted for pre-test levels of these constructs. Where baseline levels were taken into account, only one study revealed a significantly lower level of depression in non-carriers compared to carriers two years post-test (Larsson et al., 2006). No significant differences in levels of suicidal thoughts or behaviour were seen between carriers and non-carriers (Larsson et al., 2006; Wahlin et al., 2000). Whilst one paper reported levels of self-injurious behaviours and suicidal thoughts of insufficient magnitude to complete analysis (Larsson et al., 2006), another reported elevated levels for both carriers and non-carriers (Wahlin et al., 2000).

Six of the eight studies comparing the psychological impact of predictive genetic testing on carriers and non-carriers, excluded symptomatic carriers at follow-up in an attempt to obtain a homogenous group free from bias.
Understandably this undermines representative sampling and reduces the generalizability of findings. Horowitz *et al.* (2001) compared non-carriers with both asymptomatic and symptomatic carriers finding a significant difference in the levels of distress one year post-test. Symptomatic carriers reported significantly higher levels of distress than non-carriers but no significant difference between asymptomatic carriers and non-carriers was evident, which may suggest psychological difficulties are associated with disease manifestation.

Three studies revealed no clinically significant levels for either carrier or non-carrier groups on measures of psychological impact (Larsson *et al.*, 2006; Licklederer *et al.*, 2008; Wahlin *et al.*, 2000). Clinically significant levels of depression were only evident for symptomatic carrier groups (Licklederer *et al.*, 2008), or when a lower cut-off was used for the BDI (Gargiulo *et al.*, 2009).
Table II. Studies exploring differences in psychological impact of genetic testing for HD between carriers and non-carriers

<table>
<thead>
<tr>
<th>Author</th>
<th>Length of Follow Up</th>
<th>Construct Measured</th>
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<tbody>
<tr>
<td>Decruyenaere et al., 1996</td>
<td>1yr</td>
<td>BDI, STAI, MMPI</td>
<td>NS</td>
<td>NS</td>
<td>NS</td>
</tr>
<tr>
<td>Gargiulo et al., 2009</td>
<td>3m – 9yr</td>
<td>BDI, BHS, STAI, IES</td>
<td>Sig less frequent</td>
<td>Sig more frequent</td>
<td>Sig more frequent</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Sig lower</td>
<td>Sig higher</td>
<td>Sig higher</td>
</tr>
<tr>
<td>Horowitz et al., 2001</td>
<td>1yr</td>
<td>IES, BDI</td>
<td>Sig lower</td>
<td>NS</td>
<td>Sig higher</td>
</tr>
<tr>
<td>Larsson et al., 2006</td>
<td>2yr</td>
<td>BDI</td>
<td>Sig lower</td>
<td>Sig higher</td>
<td>NS</td>
</tr>
<tr>
<td>Lickledeerer et al., 2008</td>
<td>Baseline</td>
<td>BDI, GSI, SF-12</td>
<td>Lower&lt;sup&gt;a&lt;/sup&gt;</td>
<td>Lower&lt;sup&gt;a&lt;/sup&gt;</td>
<td>Higher&lt;sup&gt;a&lt;/sup&gt;</td>
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<td></td>
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<td>Higher&lt;sup&gt;a&lt;/sup&gt;</td>
<td>Higher&lt;sup&gt;a&lt;/sup&gt;</td>
<td>Higher&lt;sup&gt;a&lt;/sup&gt;</td>
</tr>
<tr>
<td>Timman et al., 2004</td>
<td>7yr – 10yr</td>
<td>BHS, IES - Intru - Avoid</td>
<td>NS</td>
<td>NS</td>
<td>NS</td>
</tr>
<tr>
<td>Wahlin et al., 2000</td>
<td>2yr</td>
<td>GHQ-30, BDI, SIBS</td>
<td>NS</td>
<td>NS</td>
<td>NS</td>
</tr>
<tr>
<td>Witjes-Ané et al., 2002</td>
<td>18m</td>
<td>UHDRS - depr - estm - aggr - com</td>
<td>Sig lower</td>
<td>Sig higher</td>
<td>Sig higher</td>
</tr>
</tbody>
</table>

<sup>a</sup> Statistical comparisons not completed and therefore cannot be determined whether statistically significant difference m: month; yr: year; NS: not significant; Sig: significance to the level p<0.05
BDI: Beck Depression Scale; STAI: State Trait Anxiety Inventory; MMPI: Minnesota Multiphasic Personality Inventory; BHS: Beck Hopelessness Scale; IES: Impact of Events Scale; GSI: Global Severity Index; SF-12: Short Form-12; GHQ-30: General Health Questionnaire-30; SIBS: Self Injurious Behaviour Scale; UHDRS: Unified Huntington’s Disease Rating Scale; intru: intrusion; avoid: avoidance; depr: depression; estm: low self-esteem; aggr; aggressive behaviour; com: compulsions

3.3 Psychological Impact of HD Genetic Test Over Time

The differential courses of psychological impact associated with genetic test results was explored in three studies (Decruyenaere et al., 1996; Timman et al., 2004; Witjes-Ané et al., 2002) presented in Table III. No study reported a significant change in the extent of depression, anxiety, low self-esteem or scored ego strength on standardised tests for carriers.
over time. After an initial peak in hopelessness after the test, carriers’ scores showed a general decrease from one year onwards. Immediately following testing, scores highlighted an increase in distress with an escalation in thoughts and feelings associated with intrusion and avoidance. Symptoms generally declined from one month post-test, although there appeared to be a peak between one and three years post-test, with avoidance being reported to a higher degree. Scores in aggressive behaviours and obsessions were seen to increase in carriers, irrespective of motor symptoms.

Non-carriers appeared to experience a decrease in depression and anxiety one year post-test and in hopelessness up to ten years post-test, compared to baseline scores. Aggressive behaviours and irritability were seen to improve with increasing time from the test result. The pattern of distress from baseline, repeatedly measured up to ten years post-test, matched the fluctuating course described for carriers.
Table III. Studies exploring changes in psychological measures following genetic testing for HD in carriers and non-carriers

<table>
<thead>
<tr>
<th>Author</th>
<th>Constructs Measured</th>
<th>Test Result</th>
<th>Findings</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>Baseline – 1m</td>
</tr>
<tr>
<td>Decruyenaere et al., 1996</td>
<td>BDI, STAI, MMPI</td>
<td>Carrier</td>
<td>No change</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Non-Carrier</td>
<td>Decreased</td>
</tr>
<tr>
<td>Timman et al., 2004</td>
<td>BHS, IES, Intru, Avoid</td>
<td>Carrier</td>
<td>Increased</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Non-Carrier</td>
<td>Decreased</td>
</tr>
<tr>
<td>Witjes-Ané et al., 2002</td>
<td>UHDRS depr, estm, aggr, irrit</td>
<td>Carrier</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Non-Carrier</td>
<td></td>
</tr>
</tbody>
</table>

m: month; yr: year
BDI: Beck Depression Inventory; STAI: State Trait Anxiety Inventory; MMPI: Minnesota Multiphasic Personality Inventory; BHS: Beck Hopelessness Scale; IES: Impact of Events Scale; UHDRS: Unified Huntington’s Disease Rating Scale; intru: intrusion; avoid: avoidance; depr: depression; estm: low self-esteem; aggr: aggressive behaviour; obs: obsessions; irrit: irritability

3.4 Non-Specific Predictors of Psychological Impact of HD Genetic Testing

Seven studies explored a range of predictors of psychological impact of testing for HD. Numerous psychological factors were highlighted, including; a prior history of symptoms in the self or family (Decruyenaere et al., 1996; Gargiulo et al., 2009; Larsson et al., 2006; Witjes-Ané et al., 2002), expectation of test result and the result itself (Gargiulo et al., 2009; Horowitz et al., 2001; Licklederer et al., 2008).
Non-specific predictors of psychological impact of HD genetic testing were identified via regression analysis (Decruyenaere et al., 1996; Larsson et al., 2006), factor analysis (Gargiulo et al., 2009) or correlation coefficients (Witjes-Ané et al., 2002). All methods are limited in their ability to infer causation and incompleteness in fully explaining the psychological impact of predictive genetic testing.

### 3.5 Methodological Issues

All the studies within this review acknowledged potential sampling bias, through examining self-selecting participants who actively sought predictive genetic testing. No studies in the current review established a difference in the psychological status of individuals at-risk of HD but who declined to undergo genetic testing. A single study discussed three sibling pairs across carrier and non-carrier groups within the sample (Wahlin et al., 2000) which may have created bias in potential survivor guilt in the non-carrier sibling.

Drop-out rates across studies varied markedly, ranging from 5% (Larsson et al., 2006) to 69% (Timman et al., 2004). Of the eight papers, only Timman et al. (2004) undertook analysis of attrition rates, reporting individuals found to be carriers who subsequently withdrew from follow-up had significantly higher hopelessness, intrusion and avoidance scores and
significantly lower psychological well-being scores than carriers who completed follow-ups, perhaps suggestive of an unrepresentative sample.

Only one study utilised a HD-specific measure with the use of the UHDRS (Witjes-Ané et al., 2002). All other measures used were standardised generic measures of psychological constructs found in the general population. Measures appeared to be used in a pragmatic manner to assess impact, rather than explicitly deriving measures from a theoretical understanding of how genetic testing might have an effect.

Since HD is a relatively uncommon disease, study sample sizes tended to be small despite reasonably large sampling frames. Consequently many reported a lack of external validity and power in finding an effect (Decruyenaere et al., 1996; Horowitz et al., 2001; Larsson et al., 2006; Licklederer et al., 2008; Wahlin et al., 2000). Five studies offered individuals tested by linkage analysis repeat testing with mutation analysis prior to their participation (Decruyenaere et al., 1996; Larsson et al., 2006; Timman et al., 2004; Wahlin et al., 2000; Witjes-Ané et al., 2002). Although not acknowledged by the authors, this may have created bias by increasing psychological burden through being genetically tested twice.
4. Discussion

This systematic review revealed eight articles that have conducted a focused examination of the psychological impact of pre-symptomatic predictive genetic testing for HD using mutation analysis. No significant differences were seen with regard to psychological impact determined by test outcome. No immediate or detrimental effects were found for individuals identified as non-carriers, rather there was some evidence of a positive psychological impact compared to carriers, albeit not reaching statistical significance. Only once symptomatic carriers were isolated from asymptomatic carriers for analysis was a significant difference in levels of distress found in comparison to non-carriers (Licklederer et al., 2008). This may be suggestive of distress being a potential early manifestation of the disease.

The course of psychological impact following testing was investigated with evidence of no significant change over time for carriers, with the exception of an initial increase in levels of hopelessness. Behavioural changes were identified with an increase in aggression and obsessions, independent of the presence of motor symptoms, which is supportive of previous research suggesting that mood and behavioural difficulties precede neurological symptoms (Folstein, 1989). Levels of depression, anxiety and
hopelessness were seen to decrease in non-carriers as well as a reduction in aggressive behaviour and irritability.

Distress (intrusion and avoidance) was seen to have a fluctuating course over time for both carriers and non-carriers, albeit at a higher level for carriers. This could be as a result of nearing the age of onset of HD for carriers (Timman et al., 2004) with theory suggesting increased distress arising from fatalism (Senior et al., 1999). Survivor guilt, regret of life choices, adaptation to result, denial of result and living within a HD family may all be involved in fluctuating levels of distress in non-carriers (Gargiulo et al., 2009).

As reported in other reviews (Broadstock et al., 2000), the majority of included studies explored the psychological impact of predictive genetic testing using general measures of affect and emotion; three of the eight appraised in this review employed a measure of behaviour and/or cognition (Larsson et al., 2006; Wahlin et al., 2000; Witjes-Ané et al., 2002). Further research including these measures would allow greater understanding of the full psychological impact of predictive genetic testing on individuals, particularly with regards to the conflicting reports of suicidal ideation reported in two of the reviewed papers.
In all but one of the studies (Gargiulo et al., 2009), clinically significant levels of the constructs under scrutiny were not found. Yet, more detailed qualitative narratives investigating individuals at-risk of HD undergoing predictive genetic testing report distressing emotions such as shock, fear and frustration at both the point of awareness about at-risk status and again with test result (Schwartz, 2010). This raises the question as to whether the measures used were sufficiently nuanced to phenomena or sensitive enough to detect emotional reactions reported by individuals. The lack of clinically significant findings may represent an absence of distress, but may also be an artefact of poor fit of measures.

Illness representations have been argued to be central in how an individual perceives their own physical and mental health, therefore playing a significant role in psychological distress (Arran et al., 2014; Rozema et al., 2009). Models of health psychology and illness representations may be usefully drawn upon to guide future research in developing and applying disease-specific, health-related measures of psychological impact. Such measures are argued to be more sensitive for the detection and quantification of clinically significant changes (Patrick & Deyo, 1989). Health-related measures such as the Health Orientation Scale (HOS, Snell et al., 1991) have been used to assess the psychological impact of being identified as a genetic carrier of familial adenomatous polyposis (FAP, Michie et al., 2001), and disease-specific measures are currently being developed and used to explore the psychological impact of cancer.
(Zebrack et al., 2006; 2008). Measures such as the Huntington’s Disease Quality of Life questionnaire (HDQoL, Hocaoglu et al., 2012) have attempted to bridge this gap for HD and it is hoped that research will utilise these measures in the future.

Three studies utilised the IES, a standard measure of trauma, and appeared to anticipate the experience of receiving predictive genetic test results as potentially harrowing, although this was not explicitly expressed by the authors. This may be an understandable orientation for research when outcomes of testing are devastating. Traumatic events are believed to shatter the assumptions an individual holds about themselves, others and the world causing them distress (Janoff-Bulman, 1992). However, from these shattered assumptions personal growth can be achieved through how we relate to others, our sense of self and our life philosophy (Tedeschi & Calhoun, 2004).

Recent evidence suggests such post-traumatic growth can occur following predictive genetic testing for HD, with both carriers and non-carriers reporting greater appreciation for life and enriched relationships (O’Rourke, 2011). Such evidence may mean that assumptions about the adverse psychological impact of predictive genetic testing should be modified, and that more theoretically robust studies, employing measures derived from constructs reflecting growth as well as pathology are warranted. Should
future research findings point to a post-traumatic growth response following predictive genetic testing for HD, there may be implications for clinical practice. Evidence from the trauma literature is suggestive of early interventions having negative consequences in psychological well-being (Bisson et al., 1997; Hobbs et al., 1996) whilst later more structured psychological approaches have reduced levels of distress (McNally et al., 2003). This may therefore be suggestive of a need for predictive genetic testing protocols to include monitoring of psychological impact over time with structured approaches offered with the identification of heightened distress.

The current review is suggestive of little psychological impact of predictive genetic testing irrespective of test outcome and may imply that living as a HD gene carrier is not in itself clinically distressing. Qualitative data elicited from HD carriers would offer some support. Identification of carrier status can generate some emotional perturbation and unwanted experiences of decisional regret and adaptation to life and goals. Yet some positive consequences such as greater life appreciation and an appreciation, and increased closeness, of family relationships have been reported (Hagberg et al., 2011). Clinically, staff who are working with patients, as they discuss options for testing, may frame discussions more positively if reassured that those who request testing do not inevitably experience adverse psychological impact. This is reflected in the most recent recommendations made to the international guidelines for predictive genetic testing in HD.
with a focus on individual choice for taking the test (MacLeod et al., 2012). However, greater consideration of how this would be introduced is warranted, given that concerns have been expressed that non-directive and full disclosure of genetic status for all at-risk individuals is both unrealistic and unwanted (Geller & Holtzman, 1995). A much larger cadre of research understanding the psychological robustness of those choosing not to undergo predictive genetic testing is needed before detailed recommendations are made.

Individuals requesting genetic testing for HD have long been described in the literature as a self-selecting group with resources and coping skills exceeding those who decline (Kessler, 1994). All current reviewed studies have utilised a self-selected sample with potentially biased findings. Indeed, genetic testing protocols and procedures have been recognized to deter all but the most motivated and determined at-risk individuals (Kessler, 1994). Research has suggested that non-testers may demonstrate higher levels of pessimism relating to themselves and their future before testing (van der Steenstraten et al., 1994) and this may serve as a self-fulfilling prophecy causing harm when unfavourable test results are obtained (Konrad, 2003). For other genetic conditions, those who electively remain ignorant of their carrier status appear to benefit from a protective function (Fanos & Johnson, 1995). Furthermore, high drop-out levels recorded in the studies reviewed herein have demonstrated higher
levels of hopelessness, intrusion and avoidance, and lower levels of general well-being in non-completing carriers (Timman et al., 2004).

All such findings suggest that greater exploration of both resilience of individuals requesting predictive genetic testing and those opting against it is needed. Achieving consensus on the most appropriate and effective protocols to address psychological impact, support individuals and their families prior to any recommendations for encouraged testing, needs additional research. This should ensure that sampling procedures will need to be adapted potentially utilising respondent-driven sampling methods or survey style questionnaires via the means of non-health related media to target the hard-to-reach population.

This is the first review of the psychological impact of predictive genetic testing by mutation analysis on individuals at-risk of HD which has employed systematic search processes. The eligibility criteria utilised permitted the reduction of bias evident in the published literature via mixed genetic testing procedures, allowing the results of the current review to be more salient and generalizable to those currently undergoing predictive genetic testing.
In conducting this review it is acknowledged that predictive genetic testing may have impacts beyond the individual undergoing testing, notably the family system (Sobel & Cowen, 2000). However, for the purpose of this review, consideration of the broader psychosocial impacts would have reduced a focused scrutiny of how the individual being tested is affected given the much larger literature that would be elicited. Given theoretical precedents for an emphasis on individual appraisal of testing and results, a focus at the individual level was felt to be warranted. Clearly broader psychosocial impacts could form the basis of a further review.
References


* Denotes references which form the basis of the current review
Part 2

Research Report

Experiences of Caring for a Partner Diagnosed with Huntington’s Disease: An Interpretative Phenomenological Analysis
Abstract

Huntington’s disease is a neurodegenerative condition conferring significant disability and requiring increasing care throughout disease trajectory. For many living with the disease, care is provided by family members, primarily partners, and for other neurodegenerative conditions research reveals such familial caring can adversely affect the carers’ psychological well-being. The partner relationship has been argued as the most sensitive to this impact.

There is limited circumscribed evidence of the impact of caring for a partner with HD, and what published literature is available is constrained by its chosen methodology. This research therefore aims to complement and add to the current literature base exploring the experiences of partner carers of individuals diagnosed with HD and explore any psychological impact of the caring process.

Qualitative interviews with six partner carers were undertaken, analysed and interpreted using Interpretative Phenomenological Analysis (IPA). Emergent themes comprised: Collective vs. Individual Care; The Invisible Partner; The Emotional Roundabout; Struggling in the Present; and Ways of Being. Sub-themes provided exploration of idiosyncrasies between respondents within broader super-ordinate themes.

All themes privileged challenges, which affirmed and disempowered those in caring roles. Dilemmas posed by responses and choices made by respondents are discussed alongside maintenance cycles of emotional phenomena identified. Themes resonated with those described by other carers with isolation, absence of reciprocity and impact on wider family members prominent. Potential clinical and research implications are discussed considering need to further explore these phenomena.
1. **Introduction**

Huntington’s disease (HD) is an autosomal dominant, inherited genetic mutation of chromosome four, estimated to affect one person per 10,000 in most European populations (Evans et al., 2013). Underpinning the disease is the expansion of the gene involved in the production of the protein huntingtin, resulting in progressive nerve cell damage in the basal ganglia and cerebral cortex, and the generation of a combination of motor, cognitive and behavioural changes. The profound impact of HD and associated decline and disability may require care across many aspects of life for those affected. Prior to clinical diagnosis, individuals with HD and their families may be hypervigilant to symptom onset and experience difficulties with changes to mood and personality including irritability and anxiety. Support may be required in completing daily household tasks, such as budgeting and driving, as a consequence of emotional difficulties and cognitive decline seen in the early stages of the disease. As the disease progresses and in later stages, care may take the form of more physically demanding tasks including dressing, feeding and toileting as symptoms progress to include dystonia (Quarrell, 2008).

As with other chronic degenerative conditions, those with HD are often reliant upon informal care. Many of those living with HD are looked after by familial carers, those of any age providing unpaid support to family members, and would be unable to manage independently (Princess Royal Trust for Carers, 2012). Many familial carers do not construct themselves
as having a formal role of care, rather extending their responsibilities as loved ones. A reluctance to label themselves as carers may contribute to an underestimate of the already acknowledged six million unpaid carers in the UK (UK Parliament, 2012) judged to save the UK economy as much as £87 billion per year.

Whilst the contributions of familial carers may well reduce the necessity of residential care, there are personal costs, with as many as 80% of carers reporting detriment to their own health and well-being (Carers Trust, 2012). Given such impacts, the need to ensure that carers themselves can continue in such valuable roles has been recognised with fundamental revisions to The Carers and Disabled Children Act 2000 and Carers Equal Opportunities Act 2004 to assure support for carers with the provision of direct services and assessments of needs (Department of Health, 2009).

However, irrespective of government aspirations, the role of carer for those living with neurodegenerative conditions confers increased risk for a number of adverse psychosocial outcomes. Research exploring the impact of caring for those with MS, Parkinson’s disease and dementias, has documented reduced quality of life and opportunities to pursue personal interests (Cummins, 2001; Ory et al., 1999) and increased social isolation (O’Reilly et al., 1996). Studies have also noted increased incidence of physical and mental health problems (Brodaty & Hadzi-Pavlovic, 1990; Figved et al., 2007; Forbes et al., 2007) with as many as 80% of familial carers of people with dementia reporting chronic fatigue, depression or
anger (Rabins et al., 1982). Caring may also enable development of personal growth and the strengthening of some familial relationships (Peacock et al., 2010). However, the weight of evidence suggests adversity as a dominant consequence of caring, with potential ramifications for well-being of carer and care recipient, and capacity to sustain care (Yeuh-Feny Lu & Guerriero-Austram, 2005).

The degree or likelihood of psychological health being affected by the caring role has been shown to be related to the relationship between the carer and the impaired person (Lawton et al., 1991). Caring for a partner has been argued to be the most intensive familial care relationship for both male and female carers with partner carers reporting greater levels of stress and depression (Pinquart & Sörensen, 2003). Within neurodegenerative conditions, greater levels of clinical depression and physical health complaints have been found in partner carers of individuals with dementia (Baumgart et al., 1992; Dura et al., 1991).

The mechanisms for carer burden have been suggested in models in which distress arises from the interplay of socioeconomic factors, personal resources and exposure to stressors during the course of caring (Pearlin et al., 1990). The physical and psychological health outcomes are influenced by objective and primary stressors (events and actions directly related to providing care), secondary role strain stressors (roles and stressors outside of caregiving) and secondary intrapsychic strains
(internal psychological processes). At each stage of the process, background and context factors influence the stress experienced. Coping strategies employed and social support networks mediate the effects of stressors and help promote positive outcomes.

Yet whilst carer response to other neurodegenerative conditions has been documented, this is far less evident for those living with HD. This is despite evidence of a greater impact of the caring role on mood and quality of life in comparison to other degenerative conditions (McCabe et al., 2009). Those few quantitative studies examining carer experience in HD have been largely assumptive and informed by models of burden, examining a broad range of family members. Studies have revealed particular concerns relating to role changes, isolation and concerns for children (Williams et al., 2012), however acknowledged that the relationship between caring and its impact was only partially examined, notably with end of life issues (Roscoe et al., 2009).

There have been attempts to capture the experience of care via qualitative methods. This has focused on familial caregiver partner dyads (Maxted et al., 2013), or largely explored specific elements of care or needs at specific time points, notably experiences of emotions (Williams et al., 2009), pre-clinical diagnosis (Williams et al., 2007), communication (Hartelius et al., 2010), and coping strategies (Soltysiak et al., 2008). In a study of impact on family caregivers, focus groups were used to examine
quality of life (Aubeeluck et al., 2012). However, the use of this method in one geographical region means carers of a relatively rare condition were likely to know one another and to raise similar issues rather than more personal experiences of care (Williams & Ayres, 2007).

1.1 Rationale and Aims of the Current Research

Caring for a partner has been argued to be the most intensive familial care relationship (Hirst, 2005), reported to increase the risk of adverse psychological effects (Dura et al., 1991) and most detrimental during transitions and key turning points in care (Burton et al., 2003; Nieboer et al., 1998). Due to the age of HD onset in middle adulthood, a partner is most likely to be the primary carer for those with the disease and for these reasons a focus on the partner relationship within this research was deemed most appropriate to understand the experience of familial caring.

The circumscribed research to date has also been constrained by an assumptive quantitative focus. Some exploratory qualitative research has explored familial carers’ experiences at significant points in HD illness, but no research to date has examined experiences throughout the disease course. This research therefore aims to understand the experiences of partner carers of individuals with HD throughout the disease trajectory, from pre- to post-clinical diagnosis of the disease.
Qualitative research to date has also largely utilised dyadic and group interviews which have merits, but may engender bias and preclude greater candour and honesty that can be afforded in individual assessment. This research hopes to complement and add to the existing literature base by using individual semi-structured interviews to offer a secure and trusting environment, allowing in depth exploration of the uniqueness and complexity of respondents’ experiences. By understanding these experiences it is hoped to supplement the evidence base to guide future guidelines and interventions for familial carers of individuals with HD.

An additional focus will be to develop an understanding of the psychological impacts of providing care for a partner with HD.
2. Method

2.1 Design

Given the lack of research exploring the partner experience of caring for those with HD, the design was chosen to generate rich data. An idiographic approach was felt most appropriate and thus individual interviews with potential respondents were designed to facilitate an in-depth understanding of the experiences of caring for a partner diagnosed with HD and any impact of the caregiving process on their lives. In attempting to understand respondents’ lived experiences an Interpretative Phenomenological Analysis (IPA) approach was employed (Smith et al., 2009).

2.2 Position of the Researcher

This research was conducted in line with the epistemological position of the researcher, namely a critical realist approach (Appendix F).

2.3 Research Context

The research was undertaken within a community service for those living with HD located across two counties in the Midlands, UK. Comprising staff from community nursing, speech and language therapy, physiotherapy, dietetics, psychiatry and clinical psychology, the team aims to provide a
comprehensive service addressing medical and psychosocial needs for those living with HD and their families. Contact and support is offered from at-risk status through all disease stages until death. Referral to the service can be expedited through a number of pathways including from themselves, genetics, neurology, psychiatry and primary care. The service provides assessment and treatment for individuals affected by HD and their families through a small inpatient unit and wider community network. Treatments offered include psychological therapy for mood and behavioural difficulties, psychiatric medication, swallowing and choking assessments as well as nursing based interventions.

2.4 Respondents

To enable in-depth analysis and development of meaningful themes both within and between respondents, the sample size for the research anticipated three to six respondents (Smith et al., 2009). This sample size allowed for identification of potentially subtle inflections of meaning (Collins & Nicolson, 2002) whilst ensuring sufficient data to perform micro-analysis of similarities and differences across cases (Smith et al., 2009).

2.4.1 Inclusion/Exclusion Criteria

Respondents were purposefully sampled from partners of current service-users of the HD service. Individuals were eligible to participate if they were currently caring for a partner with diagnosed clinical HD and had been
caring for their partner in the community for at least two years since clinical diagnosis. Respondents were required to be over the age of 18 and to speak English to a level able to participate in the semi-structured interview.

2.4.2 Final Sample

The final sample comprised six respondents, with equal numbers of males and females. All respondents were heterosexual and married. Whilst five respondents had children, none had current dependents. Respondents’ ages ranged between 55 and 80 years and all were retired. Limited demographic information is provided to maintain respondent anonymity.

2.5 Materials

Research materials used are presented in Appendix G - J including a participant information sheet (PIS), consent form, demographic questionnaire and semi-structured interview guide. Further information and description of their use is given in the following sections.

The Hospital Anxiety and Depression Scale (HADS, Zigmond & Snaith, 1983) was used as a brief self-report measure of respondents’ current psychological status prior to interview. The HADS consists of 14 items designed to screen distinct dimensions of anxiety and depression which respondents are asked to rate on a four point Likert scale. The HADS has
good psychometric properties for clinical and non-clinical samples (Bedford et al., 1997; Caci et al., 2003) and has been deemed a suitable screening tool for use with familial carers of individuals with terminal conditions (Gough & Hudson, 2009).

2.6 Procedures

2.6.1 Ethical Considerations

The research protocol was first peer reviewed by the University of Leicester academic staff and by the service-user reference group prior to seeking full ethical approval from a Local Research Ethics Committee (LREC). A favourable ethical opinion was granted following minor amendments to inclusion criteria, the participant information sheet and consent form. Correspondence to and from the LREC is provided in Appendix K. Approval was also granted from the relevant NHS trust Research and Development departments.

The PIS indicated that respondents may become distressed or upset through talking about their experiences, and if so, opportunities were provided for respondents to discuss this further with the researcher. Whilst no respondent expressed negative reactions to the interview process explicitly, two respondents revealed clinical levels of anxiety on the HADS questionnaire. Both of these respondents were currently receiving support from the host service and no further referrals to support agencies were sought.
All respondent information was stored confidentially in accordance with University of Leicester regulations. Respondents were informed both within the PIS and by the researcher directly that should they at any point during the interview disclose information which caused the researcher to believe either the respondent or a vulnerable other was at serious risk, confidentiality would need to be broken in line with the researcher’s professional responsibilities.

2.6.2 Recruitment

Recruitment of respondents was conducted between April and October 2013 and was achieved via initial discussions of its focus and remit at a carer’s group run by the HD service. In addition, discussions were held with partner carers not in receipt of carer services during nurse-led sessions in the community. Interested parties were provided with PISs and provided informal verbal consent to community nurses for their contact telephone details to be provided to the researcher with no other information about the carer or the HD service-user disclosed. The researcher then made initial telephone contact, providing respondents with an opportunity to ask questions and for an interview date to be arranged. Six individuals who met inclusion criteria expressed an initial interest in participating in the research and all six proceeded to interviews.
2.6.3 Conducting Qualitative Interviews

Semi-structured interviews are argued to be the exemplary method for IPA research (Smith & Osborn, 2003) and fit with the method’s emphasis on obtaining a detailed exploration of personal experience (Smith, 2004). Interview schedules were developed in collaboration with the HD service team to ensure they provided opportunity for respondents to discuss all elements of their experience commonly reported to staff.

All interviews were conducted in respondent’s homes, with one exception being conducted in an adult mental health out-patient department at the respondent’s request. Interviews complied with University and HD service lone-worker and safe working practice policies, ensuring that a service colleague was aware of interview locations and times with a check-in procedure following interview completion.

Prior to interview, the researcher confirmed with respondents they had received and read the PIS and ensured all remaining questions had been answered. Written informed consent was obtained. Respondents were asked to complete a brief demographic questionnaire and the HADS to give a brief measure of their current psychological status.

The interview was conducted using a semi-structured format with a series of open-ended questions and prompts. The interview schedule was developed to allow for full exploration of the respondents’ experiences in
line with the main research aims and objectives and in line with the IPA methodology. All interviews were audio recorded and ranged in length from 37 to 67 minutes. Following each interview a reflective account was written by the researcher which was later used to inform analysis.

Respondents were provided with the opportunity to discuss the impact, if any, the interview or the research process as a whole, had had on them. During this time the researcher assessed whether undue distress was evident which may have required clinical input and therefore warranted the researcher to refer to the HD service. All respondents were asked if they would like to receive a summary report of the research on its completion, which in the event that this was wanted, respondent’s addresses were kept.

2.7 Analysis

The aims of the current research were used to guide the analytic method adopted. IPA aims to explore individual processes by which individuals make sense of their experiences (Brocki & Wearden, 2006) utilising an idiographic level of analysis concerned with exploring meaning and significance of experiences at an individual level (Larkin & Thompson, 2012). The use of IPA gives respondents a voice and makes sense of the experiences they encounter (Larkin et al., 2006). Whilst it is understood that a number of different approaches could have been utilised to analyse
the current data, IPA was most suited to the aims of understanding the lived experiences of partner carers of individuals with HD.

The appreciation of individual differences within IPA in identifying both similarities and differences between respondents made this approach preferable to more narrative analytic methods. IPA’s focus on processes over time (Brocki & Wearden, 2006); complexity and personal nature of the issue (Kay & Kingston, 2001); and novelty in terms of lack of previous research and knowledge (Smith & Osborn, 2003) also justify its use within this research.

The transcription of interview audio recordings was completed by the researcher verbatim inclusive of repetitions and grammatical errors found in everyday speech. All personal identifiable information was removed at the point of transcription to ensure respondent anonymity.

Whilst acknowledging there is no one analytic method to follow, the common processes and principles described in the literature (Smith et al., 2009) were applied whilst maintaining the flexibility of the IPA approach (Willig, 2001). This included the six stages of analysis (Smith et al., 2009) comprising line by line analysis with identification of convergent and divergent patterns and themes for individuals and subsequently across respondents’ transcripts to form a joint reflective interpretative account of the data collected (Brocki & Wearden, 2006). To ensure transparency of the analytic methods utilised, further information regarding the processes
used is given in Appendix L and an example of the initial coding for a section of transcript is given in Appendix M.

2.8 Quality Issues

Quality issues in qualitative research are distinct from those in quantitative research, with questions of reliability less applicable and inherent in offering only one interpretation of the data and making few, if any generalised claims (Yardley, 2000). Quality is based on the validity of themes and credibility of the final account (Osborn & Smith, 1998). Themes were discussed with both the research supervisor and within a peer supervision group to develop coherence and ensure plausibility.

Transparency of the approach used is privileged along with explicit articulation of the epistemological position of the researcher and a need for the researcher to identify and reflect upon their own experiences (Robson, 2004; Tong et al., 2007). For this reason a reflexivity statement (an account of the process of reflecting on the potential influences of the researcher) is provided in Appendix N as a means through which readers can evaluate the current qualitative research (Crowley, 2010).

A full chronology of the research process can be seen in Appendix O.
3. Results

Through the IPA analysis of these partner accounts, five super-ordinate themes were identified which have been used to structure the results section. Each of the five super-ordinate themes were constructed from less frequent sub-themes allowing for idiographic focus of individual respondents’ experiences (Reid et al., 2005; Smith et al., 2009). Sub-themes selected and titled were understood to reflect frequency and salience of their occurrence in respondent narratives.

Whilst the structure of the results affords easier reporting of partner experiences there is acknowledgement of the complexity, overlap and dynamism of themes. A diagrammatic representation illustrating the relationship between super-ordinate themes and sub-themes is provided in Figure 1. To ensure transparency and further support the claims made within the results, the frequency of themes across respondent’s transcripts are detailed in Appendix P.

Themes describe challenges for partner carers regarding **Collective vs. Individual Care** in which dichotomous role switching can be seen alongside a need for protection and feelings of isolation. Isolation is further considered within **The Invisible Partner** in which a carer’s identity can be
self-sacrificed and individuals feel unnoticed within and outside of the relationship. Both these themes contribute to the experience of **The Emotional Roundabout** privileged by respondents in which experiences and emotional reactions can be seen as circular and can in turn create a notion of **Struggling in the Present**, reflecting on loss and change of both past and future life. The final theme portrays means of coping following experiences and decisions made regarding care as **Ways of Being** within the partner carer role. Individual subthemes are discussed in detail in relation to the super-ordinate theme which they relate to within sections 3.1 to 3.5 below.
Figure 1: Diagrammatic Representation of Themes
3.1 Theme A: Collective vs. Individual Care

This theme endeavours to capture the dichotomous nature of responses to providing collective and individual care by respondents. The adoption of approaches to care, whether forced or chosen, is considered with regards to protection and/or isolation of the family.

HD is an incurable genetic condition and its diagnosis inevitably has serious implications for the entire family. Different approaches taken for management and difficulties associated with differing emotional reactions were disclosed. For some, strength and support could be sought and received from the family as a whole. Peter described his need to return to his family to address issues collectively and mutually since he was living away from the family at the time of his wife’s diagnosis.

Anyway we got over that, you’ve got it. So there is a reason why we need to come back because that’s where the family is. (…). Yeah we’ve had a lot of trauma but you know we seem to come through it. And like I say I’ve got two daughters who are supportive. They’re both in full time
work, come and see us when they can. Tell us they’ll do anything we want, we try not to put on them

Peter

Peter’s description of having to, ‘come through it,’ emphasises an emergent strength of a family together pulling themselves through the ‘trauma,’ of diagnosis, building on their pre-existing relationships, and thus gaining strength from the collective. Sharing the news of their partner’s diagnosis with their children and their family’s response was disclosed by another three respondents and as Julia notes, it felt imperative to respond and seek solace in solidarity with others at a time of great threat.

…I’ve got to tell the girls. So we had a very emotional phone call with both of them umm because obviously then we were fully aware that it was genetic umm. So this it started to gather pace and it because this huge huge animal that was taking us with it. (…) So that was hugely traumatic. Umm, that was I think even worse than finding out about [my husband] because at least I knew I was in control of that. But with [my daughters] I wasn’t.

Julia
In addition to the sense of threatening momentum implied by an, ‘animal,’ taking them with it, Julia also suggests dichotomous roles she holds. Switching of adopted approaches by respondents was disclosed as they assimilated and immediately managed their partner's diagnosis. Respondents reported balancing the collective implications of a genetic condition with a need to, ‘control,’ with implied sole responsibility. Peter’s quote above also demonstrates dichotomous roles, whilst reporting a desire to be within close proximity to his family for support, he did not want to ‘put on them,’ and therefore implied a sense of choice in switching from a collective to an individual approach which may allow the partner to experience feelings of control. Switching may be induced by a desire to protect children.

*I think now they, they don’t discuss it, if it comes into conversation that’s fine but I don’t think anybody brings it up into a conversation unless perhaps [my daughter] does. They obviously ask about [my husband] and they want to talk to him and stuff and ask how he is and how the latest tests are and what have you but I don’t ram it down their necks because you know it’s there and its, they don’t need to know day-to-day stuff that’s going on.

*Julia*
Julia, like other respondents, reported withholding information in an attempt to shield her children and downplay any negative day-to-day experiences. Brendan describes his dilemma of protecting children at the expense of support, by not discussing difficulties to avoid causing, ‘upset,’ yet acknowledging the, ‘sophisticated help,’ which his children can provide. Rather than overwhelm children, some respondents enable their children to raise issues. Julia allowed her daughter, who has a positive genetic carrier status, to guide conversations regarding HD, in an attempt to protect her further from becoming overwhelmed with knowledge about her known future.

*I think sometimes she properly sits there and considers things and I know sometimes when she comes home she’s watching [my husband] because she obviously knows that that’s the route she’s going to take*

*Julia*

Yet adopting sole responsibility for caring for a partner was not always relished nor accepted positively.

*I’m the wife here and that’s what I am and afraid I always will be*

*Glenda*
Both Glenda and Ruth described intense frustrations at a lack of family involvement which forces their adoption of a unique responsibility. In Ruth’s case she describes a futility and anger in her inability to engage her husband’s inattentive family.

*But sometimes you really do you know want to say to them look you need to pay more attention to what’s going on and you need to be involved a bit more. But I know it’s a pointless waste of time and the more I think about that the more angry I get with them umm and the more it upsets me then because it’s...it’s just me, there’s nobody else. I don’t think they realise how bad he really is you know...well you can’t do really when you only see him for an hour a week.*

*Ruth*

Glenda discloses the very real practical challenges of caring alone with age.

*...as you get older you know you slow down really. I mean you don’t do what you used to do so umm you know it’s*
only natural isn't it really. You know I can’t do what I did in my 20’s in my 60’s if you see what I mean. It’s a bit of a difference. Sometimes there’s a big responsibility at that age to have to take it on.

Glenda

Here, Glenda’s age, and comparison with previous capabilities, contextualises her taking on additional responsibilities when caring for her husband. Glenda reflects on her care as that of a parent looking after a ‘three year old,’ and feeling resentful for needing to adopt roles that are not age and expectation concordant.

In both Glenda and Ruth’s quotes there is a sense of resignation in undertaking roles in the absence of other family members’ involvement. For both respondents there appears a magnification of loneliness and isolation as individuals as a result of having to deal with difficulties themselves.

It’s not as if I’ve got family coming in saying oh you go into town I’ll stay with him. We don’t have that situation here. I’m an only child as well, so I’ve not got any siblings so it makes it all quite lonely at times

Glenda
Isolation is not only apparent within the immediate family but is described also as occurring via schism between immediate and the wider family as a result of HD. In an attempt to distance themselves from the stigma which may be associated with genetic illnesses, wider family members appear to ignore phenomena/symptoms as previously existing or appear unaware.

…I suspect it may have been in the family but we have no way of proving that so [my wife] may not have been the first but as far as we know she is. OK without proper evidence because when I went back to her nephew he said oh no my dad hardly moved at all. He’d began to change. Because I had to tell him that this was in the family.

_Brendan_

Brendan’s experience of this dismissal of potential HD in his brother-in-law may reflect his nephew’s denial of a threatening and stigmatising disease and a wish to ignore within the wider family. The secrecy of HD symptoms even between members of the same family contributes to the difficulties in being able to manage issues using a family approach. Difficulties within the wider family and the implied discontent at how, ‘families grow apart,’ may be suggestive of how a family living with a HD diagnosis can become
isolated collectively as well as an individual partner carer within that smaller family unit.

3.2 Theme B: The Invisible Partner

Respondents’ sense of invalidation is encapsulated within the theme of the invisible partner. Not being noticed within the relationship, from others outside the relationship and from within themselves can be seen to contribute to the dismissal of feelings and a loss of personhood.

All respondents described issues relating to their lack of acknowledged presence. This could emerge as a corollary of their partner’s disease.

There are times when it’s just like living with a shell you know and it’s, it’s very upsetting at times you know (…). It’s a bit like living on your own because I’m making all the decisions but at the same time, umm I don’t want to live on my own…

Julia
Julia’s description of, ‘living with a shell,’ conveys her partner’s loss of personhood, an outer carapace which is now vacant and unable to respond. She experiences a lack of reciprocity within the relationship in which, although living with her husband, Julia describes as unwelcome and contributing to a feeling of being alone, an experience shared by other respondents. Glenda ascribes her loneliness to the lack of shared responsibility between her and her husband.

_I think sometimes you can be lonely even if you’ve doing things. In your head you can be lonely if you know what I mean. I have to initiate everything mainly which is hard work._

_Glenda_

Glenda like others, privileges descriptions of their partner’s apathetic nature and lack of motivation further intensifying a sense of going unnoticed. Ruth powerfully relates her husband’s failure to acknowledge a usually fatal medical condition, which she had minimised until compelled to visit hospital.
…it wasn’t until I got home and it really hit me what she’d said that I could have died and it scared the life out of me and [my husband] went out and mowed the lawn.

Ruth

The power of this memory, of the precariousness of her health and her husband’s dismissal and engagement with banal activity was accompanied by tears. Ruth describes an absence of once present reciprocity which had developed in her marriage and its loss provoked anger and sadness in her. Her recognition that this was attributable to HD was not enough to eliminate the feelings of not being noticed and feeling like an, ‘unpaid housekeeper.’

…that’s what I miss more than anything the fact that he doesn’t show that he cares anymore. (...) I felt like a basket case, I felt like I’d lost the plot basically. (...) I’ll never forget that day because it upset me dreadfully

Ruth

Whilst Julia, Ruth and Glenda emphasised indifference within their relationships, only Ruth accentuated how being invisible and dismissed could also be felt in interactions with others.
…the first thing they said to me was how’s [your husband]? Which is normal, and they would then say, they then said sort of thing and how are you coping? And not many people say that to me, you know, people always ask me how [my husband] is but they never sort of say how are you?

Ruth

Ruth reflects on others’ apparent lack of empathy or understanding of her own position and this seems to magnify her sense of being seen only as a carer and not an individual with needs of her own. Ruth describes that others are, ‘only interested in [her husband] because he’s the one with the HD,’ which contributes to a sense of isolation of herself as an individual within a world of HD.

However, invisibility could also emerge from an interaction between gender and care, notably with reference to gender stereotypes. This appeared most evident in the male carer respondents. Caring seemed constructed as a female activity by all male respondents with unanticipated impacts on relationships and support.
I find it difficult now to make friends. Whereas before I had lots of friends now I haven’t got any really, not men-friends. I’ve got lots of girl-friends, like you know. I make friends but there’s a lot more women then when I was like mostly men down the pub (...). But now I make it, I find it difficult to make friends with blokes basically. I’ve got nothing to talk about to them really.

Henry

Henry, because of caring, seems less able to connect with other men because his life is dominated by care in which women figure more prominently. Henry’s status however suggests equivocal invisibility, as although he feels more noticeable as a male carer in a stereotypical female role, his masculinity is reduced rendering him invisible within a male world.

Invisibility may not only be a consequence of others’ actions, inactions or appraisals. Invisibility can be self-generated. Brendan’s invisibility and isolation seemed to emerge from his choices to, ‘give in,’ relinquishing previously enjoyed activities. He challenges gender conventions by undertaking tasks to reduce carer demands on his daughters which he feels they would see as their, ‘duty,’ as females, but that leaves him more distant from others.
As respondents described atrophy and abandonment of once enjoyed activities and experiences they themselves appeared to invalidate their own needs and desires. This can affect their sense of self, mirroring the loss of personhood seen in their partners as a result of the disease.

I was trying to be this person that wasn’t panicked about anything, that could cope with anything that life threw at me, was trying to be ahead of the game with [my husband], trying to solve problems before they even arose. And then I realised that my life was actually stopping. Things that I wanted to do and enjoyed doing I wasn’t even considering that, because it didn’t fit in with the day-to-day routine and I felt as though you know at the age of, whatever I was then 58, 59 that my life had actually stopped.

Julia

Julia’s quote highlights the self-sacrifice of personal hobbies and goals, a phenomenon which formed a narrative across all six respondents’ experiences. For Julia there was an acknowledgement that this did not result in a, ‘grey life,’ for her, with her self-sacrifice led by a huge importance placed on the, ‘happiness,’ of her husband with a sense that,
‘his needs are more important.’ Self-sacrifice for the greater cause of caring was echoed by Brendan in his need to correct the neglect he felt he had shown his wife by previously completing his hobbies.

I would have liked to continue that or other activities obviously which I just couldn’t do now. I’d been neglecting her you know so.

Brendan

3.3 Theme C: The Emotional Roundabout

The metaphor of a roundabout was felt to describe the emotional reactions of caring for a partner with HD and respondents’ reflections of caregiving more generally. This theme attempts to encapsulate the circularity of emotions and the disease itself within the relentless circular movement of a roundabout with rides oscillating up and down within its motion.

The insinuated speed and incessant nature of a roundabout can be seen within respondents’ accounts of their emotional responses to the news of their partner’s diagnosis. A partner’s diagnosis with HD had immediate psychological impact on all the respondents. The circumstances and
contexts preceding diagnosis and the diagnosis itself are recalled as shocking and overwhelming.

Now they explained all what HD was and the serious consequences of it and they showed us out the door. And we went out of there holding hands and walked around [town] for half an hour not speaking because it’s a shock.

Peter

Peter discloses a sense of disbelief so intense it renders the couple unable to speak, and typifies others’ descriptions of emotional numbing. In a comparable situation offered by Julia, similar to the gaining of momentum on a roundabout as it takes its passengers round outside of their control, the speed and overpowering nature of the news of diagnosis and the need to allow time for the information to be processed is highlighted.

[The doctor] didn’t go into it gradually, just came straight out with it and said you have Huntington’s, there is no cure, it’s a genetic disease, umm we may be able to control it by drugs. That was the total conversation that we had with him. As you can imagine we came out of there
absolutely shell shocked. [My husband] didn’t say a word.

We went to the cafeteria in the hospital because we just had to sit and have a drink and gather ourselves together.

Julia

The cyclical routine of a roundabout with its oscillating upward and downward motions seemed to capture the frequent reflections of hopelessness within respondents’ accounts. Peter found his initial denial transformed into hopelessness with no choice but to, ‘accept,’ the diagnosis and the anticipated deterioration of his wife’s condition. Three respondents evidenced feelings of hopelessness within their accounts relating to the lack of a cure for the condition and consequently their feelings of a lack of control.

I just wish she hadn’t got it, I just wish we were like we were (…) she just wished she hadn’t got it, she wished she hadn’t got the walker, she wished she hadn’t got HD and wished she hadn’t got a pain in her knee. And sometimes she wishes she hadn’t got me because she tells me.

Peter
...obviously I wish it wasn’t [diagnosed] because of the repercussions with our children and others apart from us.

Brendan

Cyclical periods of hope, encouraging optimism appeared to generate further feelings of hopelessness in failing to bring about change illustrated with respondents describing the same emotions coming round again and again. Wishful thinking is used by the respondents, and their partners, as a coping strategy for dealing with the loss of their fantasised futures and lives as they knew them to be.

The maintenance of the cyclical movement of the emotional roundabout can be seen within the dilemma privileged by respondents of conflicting emotional reactions. Peter struggled with feelings of guilt from talking about his emotional needs yet also expressed anger when his emotional needs were not taken into account.

...I feel awful that, talking about her like this, but you can understand why I didn’t want to do it at home. I’ve only got a little bungalow, there’s no privacy and if she’s there I feel inhibited. If she hears me saying any of this, well that’s what you think of me is it? Oh you’re life’s not worth living
now is it? No some days it’s not dear. But any life’s better than no life. (…) It’s not nice, it’s not nice. I mean I’m still a human being. I still have feelings.

Peter

Peter reports a fear of expressing his emotional needs and associated avoidance of acknowledging them. The subsequent invalidation of his emotional needs by others may therefore contribute to his reported feelings of anger and hurt. The circularity of these emotional responses can be seen to co-exist within the circularity of HD itself.

I think it was just the shock finding out that you had this genetic problem in your family. And the guilt that you went, that went with that (…). So that was a huge huge sadness for me that we had actually given it to [my daughter] and I find that very difficult even now. Umm because you don’t want to ever hurt your child, your children no matter what and when you find out that you’ve given something that’s going to affect them so so much in later life that’s a very difficult thing to come to terms with. I mean we have but even now this, this sorrow that I feel because, I just want to make it better and I can’t. (…) But she has got on with her
life and I’m very proud of her you know she has done quite a lot of stuff…

Julia

Fears of inevitability of HD in affecting the next generation may be an impetus to continued emotional distress. Julia describes the circularity of her feelings of guilt and sadness for passing on the disease to her daughter whilst maintaining a sense of pride in what her daughter has achieved despite her diagnosis. The injustice of the perpetuation of the disease was also expressed by Brendan.

…the children were fairly bright so it seems a shame that umm you know they have this potentially hanging over them. A disease that is incurable at the moment and can strike with devastating effect…

Brendan

This circularity mirrors that of HD as a disease with its genetic element passing through generations of the same family; a never ending impact within a family, affecting parent-child and sibling relationships. The unpredictability of the disease, ‘hanging over,’ them and the fear of what will happen adds to the emotional reaction
with uncertain questions of effect and impact which cannot be immediately answered yet continues to be lived.

### 3.4 Theme D: Struggling in the Present

The theme of struggling in the present strives to make sense of the difficulties described by respondents to live their lives in the current moment. Loss and change are considered central to this theme with respondents privileging reflections and ruminations of their lives prior to caring as well as contemplation of new roles and responsibility in relation to their old lives, and to their imagined futures.

All respondents identified changes to their lives and relationships with a sense of loss. Julia, like many of the respondents, struggled with the changes to her relationship with her husband.

…it’s so difficult when somebody that you’ve lived with all these years suddenly becomes another person so the [husband] now is not the [husband] that I married. I still love him deeply but he’s not the person that I fell in love with and it’s very difficult to come to terms with that.

*Julia*
Whilst there was some acknowledgement from Julia that relationships change, ‘as you get older,’ sudden change is attributed to HD. Losses privileged by respondents included humour, communication and activities but most could be understood with an underlying theme of lack of reciprocity within the relationship. The reciprocal nature of the once enjoyed activities, interactions and relationships was something which was grieved for by all respondents. The loss of work was particularly prominent to both Glenda and Henry.

…I think I’ve struggled because as I say I retired and was just working [in my job] and mentally stimulated and having done the languages and then wham and I took an early leave. I’ve not really got over it yet really.

Glenda

…I packed in work which I did to look after [my wife], I had, I started to dream about work. I was a bit, I wanted to go back to work if you know what I mean. I was a bit upset because I used to like my job funny enough and it took a couple of years to get over it, you know not going to work. Even now I get the urge, but not very often.

Henry
Caring for a partner with HD seemed to prevent respondents from achieving relevant life goals, and appeared associated with feelings of stagnation and negativity. The loss of the desired present with a working role, and the subsequent imposed and unwanted present with a caring role, can be seen as making living in the present difficult.

The new role of caring was seen by Henry as work, describing it as, ‘harder work than when you’re working.’ The identification with a carer role brings with it a sense of responsibility beyond what would be expected from a partner relationship highlighting the changes in the present which are difficult to accept or adjust to. It is this carer identity which seems to outweigh the husband/wife identity associated with a belief of inferior needs within the relationship and an inferred responsibility to sacrifice themselves to maintain the new relationship held between them. Whilst all respondents identified with the changes to their roles, two reported, ‘resenting,’ the changes and dislike for how their lives were now, with resistance to accept the carer identity and attempts to maintain their old partner relationship. Glenda describes how caring, ‘holds you back,’ from doing other activities, burdening her with a job she never wanted to do.

…I really do not like the label carer either, as I said to [staff member] on day one. I said oh well what’s the label? (…).
The fact that I had to become a carer I said well that’s just a label I don’t want to live it.

Glenda

The present experience of loss prompted a curiosity about ‘what if’ raising nostalgia and uncertainty. Henry questioned what his life would be like had his wife not been diagnosed with HD.

I sometimes wonder what would I, if I’d, if we’d have been working and I’d have retired what would I have been doing anyway? Would I have still been going to pubs, whereas I don’t drink or smoke now… but I don’t really (...) Yeah are we better off? Are we better off like this or, obviously we’re not. But are we better off money wise like this? I don’t know. You know it’s just questions and answers really I don’t really know.

Henry

Other respondents were grieving for the loss of their imagined future which made it difficult to live contentedly in the present.
...it's been difficult for me, umm, because of what we're missing basically. And I define, I've always wanted to do something we'd always got this plan for our lives that you know that when we could afford to stop work we were going to do this that and the other together and all of this, I resent it some days that you know we'd planned 30 odd years ago, we never had a family of our own because we wanted to retire early and do all this travelling and go further afield and everything. And that's not happened and I think you know we worked all our lives and we both worked all our lives and we've got all these plans and now it's not going to happen.

Ruth

Difficulty living in the present appeared influenced by the apparent ‘if only’ ruminations of the past, with a desire for life without the disease. The new future faced by these partners does not necessarily match that which was held by them in their beliefs prior to their partner’s diagnosis. Time to assimilate the experiences of diagnosis and caring with a new imagined future is needed during which feelings of loss and grief would be common.
3.5 Theme E: Ways of Being

This theme relates to strategies employed by respondents to allow them to deal with day-to-day living. This incorporates explicit practical coping strategies and exploring different ways of approaching life as a partner carer.

Whilst living in the present was challenging, partner carers of individuals described a number of strategies to allow them to deal with immediate issues. Julia, like most respondents describes the allocation of, ‘my time,’ a time dedicated to her own needs as a means of coping.

...if I could just have you know an hour to myself in the garden, fine (...) that’s my haven. You know I go out there and I’m in my greenhouse and I’m in my zone and that’s it, that’s my down time you know and it’s boring, you know if anybody had said to me a few years ago you would have loved your garden I would have though yeah right. But it does, it’s my time out there and I can watch things grow and it gives me a more positive outlook on life.

Julia
Julia acknowledges her own needs to be kind to herself. Peter manages his emotional reactions to his wife’s arguments since her diagnosis by arguing in his head. Having arguments in his head suggests being able to acknowledge his feelings and process his reactions without exacerbating the difficulties he encounters.

What I do now, she flares up over something that I consider is trivial. For God’s sake I’ve done 98 jobs today right and this one’s not quite and she’ll really go at me. Whereas before I’d say hang on and I’d argue with her, I say nothing. I still have the argument but it’s up here [points at head]. I say what I think up here but not for her. Because it’s not going to do any good, it’s only going to make it worse.

Peter

As a relatively rare condition the amount that is known about HD is restricted and means that even those in the medical profession may have never encountered a diagnosed individual in their practice. Practical support prior to diagnosis may therefore be difficult to access. Attempting to acquire more knowledge was also used to address present issues.
They tell me it’s an extra chromosome or something like that, you know, this genetic stuff that I don’t quite frankly understand, but then again neither do the medics at this stage fully understand it

Brendan

Yet frustration was expressed that there was such little knowledge of HD and that behaviour was judged adversely.

People knew there was something wrong with him but they didn’t know what it was. They couldn’t, they didn’t put it into a box. I forget what they said it was now. Most people thought he was a drunkard but he wasn’t, he didn’t drink much but he came across as being drunk.

Henry

Ways of being were made more difficult by others’ judgement, founded on ignorance. Choreic movements were ascribed to being a, ‘drunkard,’ which whilst was not hurtful for Henry, Julia who had experienced the same misperceptions reported as being the, ‘worst part,’ and caused feelings of, ‘hurt,’ by how others perceived her husband to be. Julia saw
this as an opportunity to learn and develop through campaigning for HD promotion.

...there’s no pamphlets about Huntington’s, there’s no information on the board (...) there should be the same pamphlets about Huntington’s as there is for Parkinson’s, for Alzheimer’s, because it’s a similar animal.

Julia

Julia perceived herself as having a role and responsibility as a partner carer to help generate information and knowledge. Searching for resources and information appeared to be an effective coping strategy, whilst others actively avoided this process

I don’t really want to see too much into the future. When they start telling you this, that and the other I would rather take a day at a time and deal with that when it happens. A lot of people want to know everything about it and then I think well no I don’t want to know too much.

Glenda
In addition to this there was a further sense from two respondents of a need to appear to be coping or being seen as useful by others outside of the relationship. The differing approaches to knowledge, development and promotion with regards to HD shows the significant individual differences between coping styles of partner carers.

*I know if I said all of this to [the team member] she would say that’s the HD, I know it’s the HD, I don’t need anybody to tell me but it doesn’t stop me from trying to make things better you know and that’s how I feel that if I give up trying to make things better then he might as well not exist basically.*

*Ruth*

Some respondents were able to cope by constructing the disease and its impact as a challenge to reveal what depths of character and resolve can be drawn on. Brendan describes his caring role as a personal challenge in which he can distinguish himself, show virtue and feel positive about caring as an achievement. The caring duties are seen as a test to the relationship that was and the depth of feelings held between man and wife.
...it's a test of the depth of your character, are you able to
in these circumstances to make life easy because I could
easily see in certain situations with others that that person
could be abused.

Brendan

Shunning of the idea of personal gain coming from others’ deterioration is implied with Brendan’s continued hypothetical questioning as to whether personal challenges should be considered positive. Personal growth was identified by other respondents as a positive aspect of caring.

I think it would be very easy to become a bit of a wimp and
umm major traumas have happened, it was very easy to
step into that role whereas now although I did have
depression quite recently, because of this, I couldn’t hide
behind it. I had to move forward and it was very umm,
enlightening that I’m not so umm, weak as I thought I was.

Julia

Julia’s recognition of not being, ‘weak,’ shows her self-reflection and ability to draw benefit from her situation. Her appraisals of her behaviour and coping strategies employed may all be factors which have contributed to
finding benefit from caring. Ruth, like three other respondents privileged a method of comparing herself and her situation with others to find benefit in what she was doing.

…there’s a whole group of HD people and you know some of them have lost their fathers, husbands (...) And you think how? You know I look at them and I think I’ve got nothing to moan about

Ruth

Social comparisons demonstrate identification with others caring for partners with HD. Ruth’s ability to cope may be determined by comparisons with others identified as similar and therefore potentially provides self-enhancement. This is shown further with Brendan’s ability to place the self and the situations within a much wider cultural and societal context.

When we look at all the other diseases and problems and even without them people murder and kill and destroy, Iraq and so on, Syria and what have you. I know they’re over there but it’s still part of the human story and religion and economics and the have and the have nots. Equality and
inequality. So we’re all in that so I think you have to take a fairly phlegmatic view and say look we’re part of it make the best of it.

Brendan
4. Discussion

This research aimed to explore the experiences of caring for a partner diagnosed with HD and the psychological impact of the caring process from the perspective of six partner carers. Utilising an IPA approach, five super-ordinate themes emerged which attempted to explain the respondents' experiences namely: Collective vs. Individual Care; The Invisible Partner; The Emotional Roundabout; Struggling in the Present; and Ways of Being.

4.1 Summary of Research Findings

Respondents discussed two approaches for providing care for a partner with HD; one collectively described and another emphasising individual agency. These have both been evidenced in previous research in which pulling together of family members helps to manage and mitigate the biological, psychological and social impacts of a diagnosis (Craig & Edwards, 1983). The family system can experience a centripetal pull in which attention is refocused inwards (Newby, 1996) and permitting socialisation to the illness (Beavers & Voeller, 1983).
Movement to another mode of responding could be triggered by a perceived need to protect the self and gain control when faced by an incurable and degenerative condition such as HD. Adopting individual driven care also seemed a response to protect others and has been shown to provide a sense of mastery for parents caring for children with sickle cell disease (Hill, 1994) and in other research in familial carers of HD (Roscoe et al., 2009). Thus the current respondents’ ability to utilise both collective and individual care responses may reflect adaptive strategies protective of mental and physical health (Taylor et al., 2000). However, there appears a dilemma in choosing to employ an individual care approach. It may be protective of children and wider family members but may also foster isolation and reduce support.

Adoption of individual care was not always chosen and when imposed induced feelings of resentment and anger. This appears consonant with a finding that a diagnosis of HD can ‘close off’ a family unit (Maxted et al., 2013) with difficulties arising hiding the nature of the condition and its future implications (Williams et al., 2007; Lowit & Van Teijlingen, 2005). Such isolation may prevent the wider family from becoming fully engaged with a collective care approach. Whilst research has shown the difficulties pushing a family member into an individual carer position, the dichotomous and concurrent nature of the collective and/or individual response has not been seen previously and is worthy of further exploration.
Invalidation and invisibility, both from within and outside the relationship, were felt acutely by all respondents. Lack of reciprocity in a previously rewarding relationship appeared very hurtful. Given its centrality in building and maintaining relationships throughout the life span (Hartup & Stevens, 1997), a reduction or absence of reciprocity confers an increased risk of burnout, loneliness, depression and marital dissatisfaction (Buunk & Schaufeli, 1999). When caring for a partner, there is an increased investment in the relationship with reduced outcomes in combination with feelings of guilt and shame on the part of the care recipient (Coyne et al., 1988). However, in a HD context, due to cognitive changes, the individual diagnosed may not acknowledge such feelings, possibly leaving the partner’s response invalidated, with the potential for interventions relating to the attribution of partner intent.

Whilst invisibility experienced within and outside of the relationship was evident, respondents said little about being unnoticed by wider family members. This may reflect the focus of this research but may also reflect respondents’ deliberate distancing to protect themselves and others. Alternatively there may be a role of typical family dynamics in that this is a reconstruction of how children or siblings would generally interact with the respondents.
The lack of reciprocity available within relationships, alongside the genetic inheritance and subsequent protection of offspring from HD related issues, therefore poses a need for a unique understanding of the idea of reciprocity within a HD context. Where a partner can obtain needed validation and who should provide this is therefore questioned. Further research is required to develop these ideas with an exploration of support networks and coping.

Within the concept of invisibility respondents emphasised a loss of personhood with marked reductions in the social and relational self as well as a minimisation of their own rights and needs. Such loss of personhood has been noted in those suffering other degenerative illnesses (Albert & Mildworf, 1989), notably dementias and with interventions bringing about improvements in quality of life (Kitwood, 1991). Evidence exploring this phenomenon within parallel carer populations has evidenced a diminution of needs and a reduction of social contacts (Cummins, 2001; Ory et al., 1999; O’Reilly et al., 1996) whilst not explicitly expressing a loss of personhood. Means by which partner carers may construct their own personal identity away from their carer role may be beneficial to explore further to help alleviate the difficulties of invisibility and loss of personhood described by respondents.

Feelings of loss of personhood could be more magnified for these respondents in relation to incongruence. Partners of individuals with HD
are taking on caring roles incongruent with an expected life stage (Thomas, 1990), and may accept caring roles to the detriment of activities prioritised at that stage, potentially magnifying the emotional experience of caring (Williams et al., 2009). In not completing current life stages, challenges are made to their sense of self and subsequent personhood.

The theme of the emotional roundabout supports previously published literature in highlighting how a diagnosis impacts emotionally on partner carers. The use of grief models such as that of Kübler-Ross (1969) can be usefully drawn upon to understand the narrative of the circularity of emotional reactions to diagnosis. Anticipatory grief can be experienced by familial carers both at diagnosis and at exacerbations of the disease (Kahn, 1990) due to past, present and future losses requiring the carer to reconstruct their identity at each stage (Pera et al., 2008). Within a HD context, similar to that of dementia, the losses and changes are multiple and ongoing meaning the carer is continuously cycling through the emotional processes involved with grief (Walker & Pomeroy, 1997).

Respondents described different strategies for managing their emotional reactions, including emotional numbing and wishful thinking. Wishful thinking is well documented within familial carers of people accessing hospice care (Willert et al., 1995) and with partner carers of individuals diagnosed with Motor Neurone Disease (Goldstein et al., 1998). Wishful thinking is seen as a means of allowing experiences and losses to be
reframed in a more positive way (Moos & Holahan, 2007) and may therefore present as protective within the circularity of a genetic disease such as HD in which all hope is gone with an anticipated inevitable death of a loved one. Whilst wishful thinking may give a means of acknowledging loss and gives permission to grieve, an inability to modify imagined futures and therefore drawing negative comparisons with the present may cause a susceptibility to adverse psychological consequences.

Respondents highlighted struggles with being in the present with frequent reflections of a life and future lost. Loss has been identified in previous literature relating to caring within a HD context (Aubeeluck et al., 2012; Hartelius et al., 2010; Maxted et al., 2013; Williams et al., 2007; Williams et al., 2009) and therefore the current results are supportive of these ideas. The need to understand the human response to non-death related losses have been emphasised (Wortman et al., 1993) with carers of individuals with MS demonstrating the caregiving process to end hopes and dreams whilst raising new concerns and challenges, causing them to halt their grief to address the immediate needs of the care recipient (Cheung & Hocking, 2004).

The respondents in this study described reflections and ruminations on the past and anticipated futures yet seemed neglectful of their present lives. Whilst this may be an artefact of the questions posed, prompting a reflective process, it may also be seen as mindlessness defensively
motivated by a refusal or avoidance to acknowledge or attend to a painful present (Brown & Ryan, 2003). Such processes of looking back and forward predict levels of distress, notably in patients at-risk of genetic types of breast cancer (Erblich et al., 2000) and may be addressed through mindfulness-based interventions aimed at reducing negative affect and enhancing vitality and coping (Grossman et al., 2004). More recent identification of mindlessness within carers of the elderly and those with neurodegenerative conditions with early evaluations of effectiveness of mindfully-oriented interventions (Epstein-Lubow et al., 2011; Franco et al., 2010; McBee, 2003; 2008) may be worthy of further exploration within a HD context.

A number of coping strategies were seen to be employed by partner carers with a distinction made between practical ways of doing and more abstract ways of thinking. Consistent with previous research (Aubeeluck et al., 2012; Soltysiak et al., 2008; Williams et al., 2007) the current findings have demonstrated individual practical strategies which respondents employed as a means of managing the unique difficulties posed to them from caring for their partner. However, the distinction with ways of thinking is a unique finding of this research in considering the cognitive element of caring for a partner.

A positivist approach can be used to understand why respondents may incorporate different ways of thinking into their experience of caring. Being
able to find benefit from the caring process has been argued to improve adaptability and efficiency in responding to future stressors (Bower et al., 2009) and therefore enhances resilience (Tudage et al., 2004).

4.2 Clinical Implications

The findings of the current research have highlighted the impact on a partner of an individual’s diagnosis and subsequent caring roles adopted, warranting their inclusion within care guidelines. The European Huntington’s Disease Network released guidelines for working with individuals with HD in February 2012. The Managed Care Network acknowledged a requirement for care to extend to whole families afflicted by HD, and recognised a need for further work to clarify needs with value placed on the inclusion of family views (Simpson & Rae, 2012). Yet no clear guidelines for assessment or intervention have been developed.

The current findings have revealed coping styles employed by partner carers and has attempted to understand how and why some of these styles are adopted and their subsequent impact. In further understanding these coping styles, interventions can be tailored to meet the needs of partner carers and their wider families. An initial pilot study of an education program designed to improve quality of life for patients and their families
has demonstrated a positive impact with regards to increased coping and support seeking as well as less psychosocial burden (A'Campo et al., 2012). More studies of the effectiveness of interventions specifically tailored to the psychological issues raised by the current findings are warranted, including interventions aimed at improving the availability of support networks and an exploration of the utility of mindfulness-based interventions.

The prominence of invisibility questions where support can be found by partner carers. The dual roles held by respondents in caring for their partners with HD, whilst also providing protection to offspring, suggests clinical implications with regards to the target of any interventions: individual and/or family. The evaluation of the utility and effectiveness of family therapy style interventions may be of benefit to encourage more collective care whilst providing additional support to all involved, negotiated with the potential for increased vulnerability with perceived imposed coping. In contrast, individual interventions may provide much needed support to partner carers experiencing isolation whilst not necessarily addressing the systemic impact of the caring process of a genetic disease. Further research to explore the respective merits and disadvantages of each approach is warranted with consideration of personal choice and individual family situations.
This and previously published research has shown the reactive nature with which familial carers employ strategies to manage crises (Lowit & Van Teijlingen, 2005; Williams et al., 2009). Whilst previous research has suggested support in anticipating future challenges, the current research would encourage the exploration of concurrent interventions which support familial carers to live in the present, e.g. mindfulness. There is also an identified need for staff to be proactive in offering support due to evidenced tendencies of partner carers to self-sacrifice.

This research has supported previous comparisons with partner carer’s of individuals diagnosed with dementia (Farran et al., 2004) whilst appreciating the unique elements caring for a partner diagnosed with HD can pose. Whilst loss of personhood has not explicitly been expressed within partner carers for dementia, guidelines and interventions detailed for this population utilising a cognitive behavioural approach may be worthy of further consideration within the context of HD partner caring (NICE, 2006) to help create an identity outside of the caring role.

The need for professional and public awareness was evident from the current findings. The struggle of familial carers in obtaining information and the impact this has on their psychological well-being has previously been evidenced (Williams et al., 2007). Prior recommendations proposed by Skirton et al. (2010) in supporting professionals outside of HD care, for example through training in primary care services and the promotion of
symptoms of HD and services available with the use of public awareness campaigns, are therefore reinforced with the current findings.

4.3 Strengths and Limitations

With the limited circumscribed research on the experiences of caring for a partner with HD, the current research has added to the literature and increased understanding of their needs and experiences. The use of qualitative methodology can be considered a significant strength in providing open access to the lived experiences of the respondents. Whilst keeping a focus of the current research questions, respondents were not restricted to discuss individual elements of their experiences, frequently with adverse effect, and therefore the research allowed reflection of both positive and negative elements of their caring experience.

Previous research has highlighted the difficulties with conducting focus groups within partner carers of individuals with HD (Williams & Ayres, 2007) which may have biased previous research findings. The use of semi-structured individual interviews is therefore a strength of the current research in helping provide a safe environment to explore personal experience (Smith, 2004).
This research was conducted from a critical realist perspective, with interpretations that form the findings considered subjective. In stating the epistemological position, providing a reflexivity statement and explicitly stating the processes by which the analysis was conducted strengthens the current findings by promoting transparency (Smith et al., 2009). Whilst the concept of inter-rater reliability was redundant, quality with regards to validity to respondent transcripts and utility to answering the research questions posed were considered throughout the research process.

Whilst every effort was made to ensure the appropriateness of the methodology with regards to the research question, there was a necessity to utilise retrospective recall. Despite allowing for a temporal element in which to measure change, retrospective recall can be biased and restricted by individuals’ abilities. Research has suggested there to be improved accuracy of recall for significant and meaningful life events (Blaxter & Paterson, 1982). This, along with the recruitment of current carers to ensure caring evoked, ‘hot cognitions,’ to recall (J. Smith, personal communication, 6 August 2012) was hoped to improve credibility of findings by respondents recalling intact cognitions. Efforts were also made to structure interviews to allow recall around key life transitions to aid with memory (Humphrey, 1993).

The emphasis of the IPA methodology is at an idiographic level of analysis, therefore interested in how individuals make sense or give
meaning to their experiences. Memories recalled by respondents are interactions between their own life narratives and societal discourses (Hunt & McHale, 2007), and therefore the telling of their stories allows for personal meaning to be explored and uncovered (Wrye, 1994). Recall of memories is likely to be altered over time and influenced by many factors (Bartlett, 1932; Burnell et al., 2006) but this is key to understanding the individual’s experience. Therefore the bias which may be created with retrospective recall is not as significant within qualitative research.

IPA requires a relatively homogenous sample to allow insight into a particular experience in a particular context (Smith et al., 2009), which provides in-depth analysis and understanding but can make few generalised claims to the wider population. The current sample encompassed both male and female partner carers in the middle stages of the disease trajectory. It had been expected with the average age of onset of HD between 35 and 55 years (Quarrell, 2008) that the sample age would have been younger. Further research to explore differences in younger partner carers may be of interest, particularly with regards to the life stages model. Further to this, the current sample consisted solely of heterosexual couples. Whilst sampling did not exclude homosexual partner carers, the accessibility of such respondents was not obtainable. It is acknowledged that this sub-group may have different or additional needs to the sample explored within the current research and further exploration of their experiences would be warranted.
### 4.4 Recommendations for Future Research

The current research has highlighted a number of areas worthy of further exploration with future research. Whilst the current findings have provided information on partner carers’ experiences of caring over the course of HD, a longitudinal study in which respondents are returned to throughout the disease trajectory would allow for improved understanding of any changes in their experience over time and subsequent coping strategies. A longitudinal approach would also place less reliance on the use of retrospective recall, the weaknesses of which have been discussed above.

Whilst qualitative research is limited in its ability to make generalised claims to the wider population, areas worthy of further exploration are highlighted to guide potential future research utilising quantitative methodology. In confirming and quantifying phenomena such as isolation, reciprocity, coping styles and strategies highlighted by the current research, recommendations for future interventions can be developed with subsequent evaluation of their effectiveness.
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Part 3

Critical Appraisal
1. Introduction

During the completion of this research, a reflective journal was kept on which this critical appraisal was based. The purpose of this paper is to allow for personal and professional reflections on the experience of completing the research as well as provide a summary of what was learned throughout the process.

2. The Research Journey

2.1 Choosing a Research Topic

Prior to training, I had very strong interests in neurodegenerative conditions and in particular dementia with experience completing research and clinical work in this area. My personal reasons for entering a career in psychology began with my grandfather’s diagnosis of vascular dementia which bought him into contact with a Clinical Psychologist. Throughout my grandfather’s disease I gained an interest in his relationships with different people and the impact of his diagnosis on their perceptions of him, and subsequently their interactions with him. This interest began to extend to the experiences of my family members as they took on caring roles. As I grew older and began to take on a caring role for my grandfather myself, I began to reflect on the differences in my experiences, both within my
relationship with my grandfather and my individual identity when caring for him. The systemic impact of neurodegenerative illnesses is what initially triggered my interests in exploring the experiences of familial carers.

On investigating the current literature base on familial carers of individuals with dementia I began to feel overwhelmed by the extent of the published research. I began to question the impact any conducted research would have in the area. It was at this time that I began to look into experiences I could obtain on placement and was introduced to services for individuals with HD. Whilst the comparisons with dementia were obvious to me from the start, in particular with regards to cognitive decline, I was shocked to find out the extent of the reliance on dementia research within the HD field. To me it seemed that the uniqueness of HD was not fully appreciated with regards to the impact of its genetic inheritance and age of onset, alongside its triad of impairment in motor function, cognition and emotion. This really provoked a determination within me to try to combine my interest in familial caring within a HD context in order to bring about interest in a somewhat neglected field.

2.2 Choosing a Methodology

Within the current research topic, I felt a qualitative approach was most appropriate given the limited amount of previous research and the
expressed need to establish family views in the development of professional guidelines for working with individuals with HD and their families (Simpson & Rae, 2012). Prior to training I had no previous experience of qualitative methodology and was keen to use the opportunity of training to develop a new skill and understanding of the benefits of this type of research.

I explored different qualitative methodologies prior to alighting on IPA. With an aim of the research being to explore lived experiences over the course of HD caring there was a requirement for the methodology to incorporate a temporal element within the analysis. The more traditional longitudinal analytic methods were unfortunately not available to me with the time restrictions of completing the research as part of the DClinPsy. I chose to contact experts of qualitative methods and felt confident that whilst other approaches would have their benefits to exploring the research question posed, IPA allowed for analysis of individual differences as well as similarities in processes over time (Brocki & Wearden, 2006). By confirming that respondents were still actively caring for their partner with HD I was able to ensure that they were reporting on ‘hot cognitions’ when recalling retrospective memories. The need for retrospective recall is obviously a limitation to the current research and with a greater time allowance I would have considered alternative methodology.
2.3 Collecting Data

I was reliant on the HD team Community Nurses in the recruitment of participants. I therefore was strategic in engaging with them to understand both the service and its priorities. Their sustained interest in research was advantageous but I came to appreciate the importance of negotiation and showing the relevance of research to stimulate goodwill. This approach seemed to be facilitative and I had no real difficulties in recruiting six participants, the upper end of my proposed sample size.

During interviews with respondents I was struck by the openness and honestly with which they let me into their lives and shared their experiences. I was touched by the need of some respondents to introduce me to their partners to allow me to further understand some of their experiences in a more personal way. This challenged my role as solely a researcher in this context. I found myself wanting to explore specific difficulties and subsequent emotional reactions to situations, and address client well-being, but was mindful not to influence the narratives provided by respondents.

On meeting respondents, issues relating to their individual experiences of caring for their partners with HD were raised with me prior to the interview. When this occurred, I emphasised to respondents that the topic seemed
very important to them and that they would be very welcome to speak about this once recording had started to allow for the information to be included within the later analysis. On several occasions, once I had provided respondents with the opportunity to raise these topics they chose not to. On one occasion the respondent chose to restart their conversation about the topic only once the recorder had been switched off. This was somewhat disappointing for me in feeling I had missed out on the opportunity to fully appreciate the whole of the partner carer experience. Through reflection I developed an understanding of this as whilst talking about some personal issues was acceptable for respondents, there was something about making a permanent voice recording of these views that somehow crossed a barrier for them with regards to their feelings of guilt and disloyalty already disclosed during interviews.

To help facilitate respondents to feel able to share information and therefore improve the credibility of findings, research has looked at providing email interviews as well as utilising other electronic mediums. By providing anonymity, email interviews can improve self-disclosure whilst also benefiting from more focused and reflectively dense accounts being obtained from respondents (Curasi, 2001; Meho & Tibbo, 2003; Murray 2004; Murray & Harrison, 2004). This however needs to be balanced with the methodological weaknesses in terms of loss of non-verbal cues which can reduce the quality of the data obtained, as well as requirements for respondents to be technologically competent (Meho, 2006). The current
research findings could have been strengthened with the incorporation of respondent diaries to provide complementary material to the more traditional semi-structured interview data (Reis, 1994) whilst also allowing for the processes of change throughout disease trajectory to be studied (Bolger et al., 2003).

In respondents choosing not to disclose elements of their experience during interview, the credibility of the subsequent findings could be questioned. The issues of missing information during interview are seen as an inherent factor of naturalistic research (Potter & Hepburn, 2005) and therefore common within qualitative methodology. On reflection, I could have explicitly asked respondents whether they would have liked me to ask them directly about these issues during interview and therefore been led by their feelings with regards to the topic which may have increased the credibility of the findings.

At times I felt irritated by respondents’ apparent inability to discuss their own reactions to situations, instead focusing their attention on the reactions of their partners and the rest of their family. During interviews I recalled trying to make my interests in them as individual’s explicit, sometimes to no avail and triggering feelings of frustration. This may be a representation of the reduced self-concept caused by the restriction of normal life activities as a result of caring (Skaff & Pearlin, 1992) and therefore in switching focus back to themselves may have highlighted
these changes and thus been too painful for them to acknowledge. On reflection after the interviews I was surprised by my annoyance at this, given my own experiences of caring prior to commencing the research and in particular the shift of focus onto the care recipient at the cost of my own identity and needs.

2.4 Transcription and Analysis

I chose to undertake transcription myself to fully immerse myself in the data and reduce the likelihood of bias in using a transcriber (Halomb & Davidson, 2006; Tilley, 2003). Having not used IPA before, I attempted to become more familiar with the methodology firstly by reading prominent texts (Smith et al., 2009) and secondly by attending an IPA workshop teaching the common processes and principles (Smith et al., 2009).

At the suggestion of my research supervisor, prior to conducting interviews, I spent some time considering my epistemological position. Throughout the analytic process I was interested to see how my interpretations were influenced by my previous experiences and world views and acknowledging that these interpretations may not necessarily be ‘true’ for others. I was conscious of wanting to remain transparent in my methodological approach and therefore chose not to have a supervisor.
counter-code my transcripts as, from a critical realist perspective, our individual codes and interpretations would be influenced by different personal perspectives and meanings (Shaw, 2010). Instead I chose to discuss my themes with my research supervisor and with a qualitative methodology peer group formed from members of my cohort. The themes were only discussed on the basis of usefulness and being a valid representation of the experiences offered within individual transcripts.

Analysis was time consuming and the amount of data generated via interview at times felt overwhelming. Analysing my first transcript took considerably more time than anticipated and I could feel tension in my need to be perfect. I countered this being mindful there was, ‘no clear wrong or right way,’ (Smith et al., 2009) to conduct IPA and returned to the individual transcript to ensure my interpretations reflected the words spoken of the respondents.

In creating interpretive themes I was concerned that I might lose the idiosyncrasies of individual respondents’ experiences and questioned whether the process was too reductionist for my personal style. However, as I continued with the method I became aware of its utility in revealing individual similarities and differences. It was both enlightening and liberating to write my results fully using respondent’s own words, and giving more personal meaning to their experiences beyond the numerical capture of a quantitative approach.
2.5 **Dissemination**

Given the often marginal status of HD as a relatively infrequent diagnosis, I felt that it was very important to share this research beyond traditional academic settings to allow for the voices and experiences of respondents to be heard. In addition to more traditional conference dissemination I will present my findings to the HD service, with management present, to allow professionals involved in planning and delivering care to individual’s diagnosed with HD and their families to better understand partner carers’ experiences. Inclusion of those in managerial positions may provide leverage for change. I hope that this will subsequently drive future service delivery to better meet their needs. I intend to provide a summary report to the HD service, as well as any respondents who had previously expressed an interest. I am also planning to present my research at a research conference to be held in September 2014.

In highlighting areas in which quantitative research can be conducted to support claims made (Cutcliffe & McKenna, 1999), which have been discussed in the research report section of this thesis, the effectiveness of the current research dissemination is hoped to be enhanced allowing for useful clinical implications to be drawn which will bridge the gap between research and clinical practice (Kazdin, 2008). Due to my current placement taking place within the HD service of which this research was conducted, there is a unique opportunity for me to implement these
findings into my clinical practice. I am intending to co-facilitate a Caring with Confidence group in Summer 2014 for families of individuals diagnosed with HD and the findings of this research will be used to help guide the subject content, discussions and information provided. Whilst my research and its findings have been welcomed by the HD team, my current trainee status has meant I am not fully embedded within the service and therefore will not be able to see the longer term impact of the results. To prevent fading of momentum with regards to research in this field, I will conduct project planning with both research and clinical supervisors to encourage further salient research to be conducted in the future.

The literature review which forms a substantial part of this thesis has already been submitted for publication in March 2014. The process of transforming a section of thesis into a journal article required the adoption of different formatting styles and therefore I faced additional demands and effort during a time when my focus was primarily on writing up for thesis. I was required to consider an alternative audience for the article with the consideration of a readership not necessarily exclusive of clinical psychologists and therefore a difference in assumed knowledge was fundamental during the writing process. I hope that this will add to the literature base regarding the psychological impact of predictive genetic testing for HD and thus be used to help at-risk individuals and their families to make more informed decisions regarding the process.
I also intend to publish the empirical research following submission of this thesis. The process of writing up sections of this thesis for journal submission will need to maintain research context and researcher reflexivity to maintain rigour and credibility (Robson, 2004; Tong et al., 2007). By focusing on key themes which are relevant and applicable to clinical practice, findings can portray a greater depth of understanding to readers, therefore increasing utility.

3. **Reflections on Professional and Personal Development**

I have learned a great deal through completing this research, both as a professional and about myself personally. I have challenged myself to utilise a method previously unfamiliar to me in terms of its epistemology and techniques. It has been enriching and confidence building and I am proud of the work which I have produced. The research process and approach has informed my clinical work. I feel better able to see systemic issues and will perhaps privilege the voice of the service users and their families in formulations where appropriate.

I have been able to fully appreciate my long standing preoccupation with organising and planning my work load and feel that these skills have been
enhanced throughout this process. Without this organisation, I feel that the amount of work involved with completing this research could easily have become overwhelming.

The new skills and knowledge of HD and the psychological impact that a disease such as this can have not only on an individual but on a whole family system is something which I will take away from this work. I have found it to intensify my interest of the psychological impact of physical health conditions and increased the personal value I place on the application of clinical psychology ideas and ways of working beyond general mental health settings.

Throughout this research process I have frequently been reminded of advice offered to me by another trainee, of the importance of, ‘self-care as a practitioner,’ (Bond, 2000). The words of this trainee, as well as subsequent clinical interests in developing self-compassion as a mean of coping (Gilbert, 2005; 2009; Neff, 2003) have been helpful to me and I truly value the importance of being able to recognise what is ‘good enough’ and developing strategies to be a little kinder to myself. Through this process I believe I have built up some of these skills which I will take forward and hopefully continue to develop further.
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Appendices
Appendix A: Guidelines for Authors for Literature Review Journal Target


Manuscript Submission

The Journal of Genetic Counseling uses a fully web-enabled online manuscript submission and review system. To keep the review time as short as possible, we request authors to submit manuscripts online to the journal's editorial office. Our online manuscript submission and review system offers authors the option to track the progress of the review process of manuscripts in real time.

Manuscripts should be submitted to: http://jogc.edmgr.com

The online manuscript submission and review system for the Journal of Genetic Counseling offers easy and straightforward log-in and submission procedures. This system supports a wide range of submission file formats:

For manuscripts — Word, WordPerfect, RTF, TXT and LaTeX; for figures — TIFF, GIF, JPEG, EPS, PPT, and Postscript. PDF is not an acceptable file format.

After a manuscript has been accepted for publication and after all revisions have been incorporated, a final manuscript should be submitted through the online submission system. The electronic file submitted must be the finalized version of the manuscript. The author may track the status of a submission via the online submission system at any time.

NOTE: If you encounter any difficulties while submitting your manuscript online, please contact the Editor-in-Chief, Bonnie LeRoy, via e-mail at: leroy001@umn.edu

General

Manuscripts should be checked for content and style (American English spelling, punctuation, and grammar; accuracy and consistency in the citation of figures, tables, and references; stylistic uniformity of entries in the References section; etc.)

Comments section: Authors should detail in the comments section of the submission that the manuscript is submitted solely to this journal and was not published elsewhere, and disclose details of any previous or
anticipated publication history related to the manuscript's content. Submission is a representation that the manuscript has not been published previously and is not currently under consideration for publication elsewhere.

**Manuscript Preparation**

1. Type double-spaced and include all illustrations and tables. Original research articles should be no longer than 25 double-spaced typed pages and qualitative research no longer than 40 double-spaced typed pages.

2. Title page: A title page is to be provided and should include the title of the article, authors name (no degrees), authors affiliation, and suggested running head. The affiliation should comprise the department, institution (usually university or company), city, and state (or nation) and should be typed as a numbered footnote to the author’s name. The suggested running head should be less than 80 characters (including spaces) and should comprise the article title or an abbreviated version thereof. The title page should also include the complete mailing address, telephone number, fax number, and e-mail address of the one author designated to review proofs.

3. Abstract: An unstructured abstract is to be provided, approximately 200 words

4. Key words: A list of 3-10 key words is to be provided directly below the abstract. Key words should express the precise content of the manuscript, as they are used for indexing purposes.

5. Section headings: All major sections should carry section headings (such as Introduction, Methods, Results, Discussion, Conclusions, etc.) type centered. Side headings in Methods section should include, as appropriate: Participants, Instrumentation, Procedures, and Data Analysis. Side headings in Discussion should include: Study Limitations, Practice Implications, and Research Recommendations. All Acknowledgements (including those for grant and financial support) should be typed in one paragraph (so-headed) on a separate page that directly precedes the References section.

6. Reference list: The journal follows the reference and citation style recommendations of the Publication Manual of the American
Psychological Association (APA style). See also: http://apastyle.apa.org/

List references alphabetically at the end of the paper. References should include (in this order): last name and initials of authors, year published, title of article, name of publication, volume number, and inclusive pages. Where there are seven or more authors, abbreviate the seventh and subsequent authors as et al.

Refer to the references in the text by name and year in parentheses. Multiple citations should be listed alphabetically by author’s last name.

7. Illustrations: Illustrations (photographs, drawings, diagrams, and charts) are to be numbered in one consecutive series of Arabic numerals. The captions for illustrations should be provided. Photographs and drawings should show high contrast. Electronic should be in TIFF or EPS format (1200 dpi for line and 300 dpi for half-tones and gray-scale art). Color art should be in the CMYK color space. A hard copy of photographs or illustrations may be requested prior to publication.

8. Tables: Tables should be numbered (with Roman numerals) and referred to by number in the text. Each table should be on a separate sheet of paper at the end of the submission. Center the title above the table, and type explanatory footnotes (indicated by superscript lowercase letters) below the table.

9. Footnotes: Footnotes should be avoided. When their use is absolutely necessary, footnotes should be numbered consecutively using Arabic numerals and should be typed at the bottom of the page to which they refer. Place a line above the footnote, so it is set off from the text. Use the appropriate superscript numeral for citation in the text.

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Conflict of Interest
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Human and Animal Rights

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If supplying any supplementary material, the text must make specific mention of the material as a citation, similar to that of figures and tables.

Refer to the supplementary files as “Online Resource”, e.g., "... as shown in the animation (Online Resource 3)", "... additional data are given in Online Resource 4”.

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For each supplementary material, please supply a concise caption describing the content of the file.
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The manuscript contains a descriptive caption for each supplementary material

Video files do not contain anything that flashes more than three times per second (so that users prone to seizures caused by such effects are not put at risk)
Appendix B: Glossary of Terms

At-risk Individual: Individuals who have a family history of HD. If a parent is known to be effected, the individual holds a 50% risk of developing the disease themselves. If a grandparent is effected (with the parents’ status being unknown), the individual holds a 25% risk of developing the disease themselves.

Autosomal dominantly inherited: Pattern of inheritance of a disease where only a single copy of the disease gene is required in order for the individual to develop the condition.

Carrier: Individual who has undergone predictive genetic testing and been given a positive result i.e. they will go on to develop the disease.

Confirmatory genetic testing: Symptomatic individual who has undergone genetic testing in order to obtain a clinical diagnosis.

Direct mutation analysis testing: Genetic testing procedure whereby the exact mutation of the DNA associated with a disease is tested for in an individual’s DNA giving up to 100% accuracy.

Intermediate alleles: Individuals with a CAG repeat length of between 27 and 35, who will not develop symptoms of the disease themselves, but their children will be at-risk of HD.
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<td>Linkage analysis testing:</td>
<td>Genetic testing procedure whereby a section of DNA is identified as having involvement in a disease and compared between individuals known to be affected and family members with an unknown status. If multiple loci of the area of DNA match there is a higher percentage change of disease inheritance.</td>
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<td>Non-carrier:</td>
<td>Individual who has undergone predictive genetic testing and been given a negative result i.e. they will not go on to develop the disease.</td>
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<td>Pre-natal genetic testing:</td>
<td>Predictive genetic testing conducted on a foetus to establish whether they will go on to develop the disease.</td>
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<tr>
<td>Reduced penetrance:</td>
<td>Individuals with a CAG repeat length of between 36 and 39 who may or may not develop symptoms of the disease.</td>
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<td>Trinucleotide:</td>
<td>A group of three compounds which make up DNA, consisting of adenine (A), thymine (T), guanine (G) or cytosine (C).</td>
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### Appendix C: Database Searches

**Scopus (07/11/2013)**

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**PsycInfo (07/11/2013)**

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**Medline (07/11/2013)**

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## Appendix D: Data Extraction Form

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</tr>
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</tr>
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<td>Toufexis &amp; Gieron-Korthals (2010)</td>
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<td>Van &amp; Ten Kroode (1997)</td>
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Appendix F: Statement of Epistemological Position

The current research was conducted from a critical realist epistemological position. This position integrates the realist ontology with relativist epistemology (Issac, 1990), in which the world and all realities within it are considered from a position between the extremes of naïve realism and extreme relativism (Willig, 2001) i.e. reality exists non-contingent on human perception (Bhaskar, 1978; 1986).

No reality can be seen as objectively knowable, with the unique interpretations and meanings individuals place on experiences creating distortion (Shaw, 2010). Respondents and researchers are intrinsically linked by a double hermeneutic cycle in which the reality and meaning of the respondent’s experience is interpreted through interaction with the researcher’s own individual view of reality (Heidegger, 1927).

A primary purpose of research is judged to be the attainment of knowledge of underlying causal mechanisms. Whilst not directly observable, the structures, powers and relations that explain phenomena beneath the surface level are considered real and identifiable through their effect (McEvoy & Richards, 2003). A relativist stance in which it is deemed not possible to make comments on the nature of an individual’s reality, due to no direct contact with it, means that interest is taken in what contact there is, i.e. what is said by the individual (Harper, 2012).
In line with this epistemological position, whilst attempting to understand the lived experiences of caring for a partner with HD, the researcher acknowledges the influence of their own view of reality in not only understanding their own experience of the research process, but also their attempt at interpreting the respondents’ accounts of their experience (Smith et al., 2009). Therefore the discussions and comments made within this research are considered as the researcher’s own interpretations of the respondents’ experiences and not as absolute truths of reality.

References:


*Journal for the Theory of Social Behaviour*, 20, 1-31


Appendix G: Participant Information Sheet
Printed on headed paper – Version 3 (18th January 2013)

Participant Information Sheet

Experiences of Partner Carers of Individuals with Huntington’s Disease
Through Pre and Post Clinical Diagnosis and the Effect of the Caregiving Process

You are being invited to take part in a research study. Before you decide whether or not you wish to participate, it is important for you to understand why the study is being done and what it will involve. Please take the time to read the following information carefully and discuss it with others if you wish. Thank you for reading this.

What is the purpose of the study?

The study aims to interview partner carers of individuals with Huntington’s disease (HD). The study is looking at experiences over the course of the disease as seen by partner carers and any effect this has had on them. The study aims to help better understand HD and the impact of caring as well as identify needs of partner carers of individuals with HD and help drive future developments for interventions and support programmes.

Why have I been chosen?

Participants for the study were identified from the HD service of *** NHS Trust. Partner carers of individuals with HD known to this service have been approached to take part. This information sheet will have been provided to you by a member of the *** HD service team and no personal details have been passed to any researcher involved in this study without your consent.

Do I have to take part?

It is up to you to decide whether or not to take part. If you decide to take part you will be given this information sheet to keep and be asked to give your written consent. If you decide to take part you are still free to withdraw up to three days after your interview without giving a reason. Any information or responses you may have already given will be destroyed. A decision not to take part or to withdraw at any time will not affect the standard of care either you or your partner will receive.

What will happen to me if I take part?

Once you have agreed to take part in the study a researcher will arrange to come to see you at your home at a mutually convenient time in order to complete an interview. Interviews taking place in your home will need to be conducted in a private place. If a suitable room is not available at your home, or you would prefer not to be interviewed at home, an interview can be conducted in a convenient NHS building. Should you choose for your interview to be conducted within an NHS building you will be reimbursed for your travel expenses to and from the interview location. You will be asked to complete a Hospital Anxiety and Depression Scale about how you have been feeling over the last week and a short demographic questionnaire prior to the interview. The interview should take no longer than an hour. You will be asked to give written consent for the interview to be audio recorded. Only one interview is required. If it is not possible to complete the interview in one appointment then the researcher will be happy to arrange to visit you again. Breaks can also be taken at any time throughout the interview.

There will be an opportunity after the interview to discuss with the researcher any questions you may have or any concerns or distress caused. The information will be

______

5 *** denotes deletion of identifiable information to ensure service anonymity
gathered to look at partner carer’s experiences over the course of the disease and any effect this may have had on them.

**What do I have to do?**

Taking part in this study means that you will be asked to complete two short questionnaires and be interviewed. You do not have to do anything else. Your regular activities and day-to-day routines will not be affected as much as possible.

**What are the possible risks or disadvantages of taking part?**

There are no risks involved in taking part but it is possible that talking about your experiences may cause you to feel upset or distressed. The researcher will therefore always offer the opportunity after the interview to discuss this with them and support will be provided if you have found it distressing in any way. A break can be asked for at any point within the interview should distress become too great.

**What are the possible benefits of taking part?**

We hope that this study will help you by discussing your experiences of caring and any perceptions you may have had over the course of caring for your partner with HD. However, this cannot be guaranteed. We are also hopeful that findings from the research may be used to help services be more sensitive to the needs of patients and their families and inform future clinical practice.

**What if something goes wrong?**

If you wish to complain or have any concerns about any aspect of the way you have been approached or treated during the course of this study, the normal National Health Service complaints mechanisms are available to you.

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

All information which is collected about you during the course of the study will be kept strictly confidential. All information recorded from you in the interview will not have any information attached to it that you could be recognised from. Interview data will be kept on encrypted computer hardware and will be destroyed five years following the study.

Should information be disclosed during the course of the research which causes the researcher to believe either you or a vulnerable other is at serious risk your confidentiality may need to be broken, in line with *** NHS Trust policy. In the event of this occurring, wherever possible the researcher will discuss that your confidentiality will be breached.

**What will happen to the results of the study?**

The results of this study will be available in 2014-2015. The results will form the researcher’s doctoral thesis and is expected to be published in a peer-reviewed journal. A copy of the collected results will be sent to the HD service and any other interested organisation. If any participants wish to receive a report about the study’s findings they can inform the researcher at the time of the interview.

**Who is organising the study**

The research is organised by the University of Leicester and *** NHS Trust. The National Research Ethics Service (NRES) Research Ethics Committee *** – *** has reviewed the study. The research has obtained NHS permission and Research and Development approval from *** NHS Trust and *** NHS Foundation Trust.
Contact for further information

Thank you for considering taking part in this study. If you have any questions or would like more information about the study please contact one of the researchers on the following numbers:

[researcher details]
Appendix H: Participant Consent Form
Printed on headed paper – Version 3 (18th January 2013)

Participant Consent Form

Experiences of Partner Carers of Individuals with Huntington's Disease Through Pre and Post Clinical Diagnosis and the Effect of the Caregiving Process

Consent Statements:

| I. | The nature of the research project has been fully explained to me and I am aware of what my participation will involve. I have been provided with a detailed participant information sheet (Version 3 – 18th January 2013) and understand that I will be required to be interviewed by a researcher. |
| II. | I have been given the opportunity to ask questions about the research and these questions have been satisfactorily answered. |
| III. | I understand that the interview will be audio recorded and transcribed with all data kept confidential in line with University regulations. I understand that audio tapes will be confidential and only listened to by specified people involved in this research project. I understand that audio tapes will be stored in a secure locked environment during the course of the research project and following completion of the research all tapes and data will be held in a locked room at the Clinical Psychology Base at the University of Leicester and destroyed after five years, in line with university regulations. I understand that there will be no identifying information on the tapes or transcripts of interviews being stored. |
| IV. | I understand that my participation is voluntary and that I may withdraw from the research up to three days post interview, without giving reason. In this instance I understand that any audio recordings will be erased and my or my partner’s future care will not be affected in any way. |
| V. | I understand that the risk in this study is that I may become upset or distressed through speaking about my experiences and that I can ask for a break in the interview or a complete termination at any time and without giving reason. |
| VI. | I understand that the research forms part of the researcher’s doctoral thesis and is intended to be published in a peer-reviewed journal. I agree for the use of direct quotations from interviews to be used within said documents with the understanding that no identifying information is included. |
| VII. | I understand and agree for study documents to be accessed by regulatory authorities including the NHS trust as necessary. |

I am submitting myself for participation in this research project with the full knowledge and understanding of the nature of the research project and of what will be expected of me. I agree to all consent statements detailed above.

Participant Signature: ________________________________
Name in Block Capitals: ________________________________

Researcher Signature: ________________________________
Name in Block Capitals: ________________________________
Appendix I: Demographic Questionnaire

Experiences of Partner Carers of Individuals with Huntington's Disease Through Pre and Post Clinical Diagnosis and the Effect of the Caregiving Process

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<th>Female</th>
<th>Prefer not to answer</th>
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<td>25 – 34</td>
<td>35 – 44</td>
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<td>2</td>
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<td>Second generation</td>
<td>Third generation</td>
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<tr>
<td>HADS score:</td>
<td>Anxiety</td>
<td>Depression</td>
<td></td>
</tr>
</tbody>
</table>
Appendix J: Semi-Structured Interview Schedule

Version 2 (15th November 2012)

1. Can you tell me about life prior to your partner receiving their diagnosis of HD?
   
   Prompt: Relationships, family life, social life, work life…

2. How would you describe yourself as a person?
   
   Prompt: What kind of person are you? What are the most important things to know about you? How do you see yourself?

3. Has having a partner with HD affected how you see yourself and if so how?
   
   Prompt: How would you say you have changed? How would you have seen yourself before your partner was diagnosed with HD?

4. Can you briefly tell me about the process of your partner receiving their diagnosis of HD?

5. Can you tell me about your life currently?
   
   Prompt: Relationships, family life, social life, work life…

6. On a day to day basis, can you talk about any changes you might have noticed in your partner?
   
   Prompt: Are there any differences in your partner as person? How do you explain any differences? – Are these differences a direct result of having HD? How do you cope with any changes?

7. How would you describe the impact on you of caring for a partner with HD?
   
   Prompt: Are there any positive aspects to caring for a partner with HD? Are there any negative aspects to caring for a partner with HD?

8. Can you tell me about how you see your life in the future?
   
   Prompt: Relationships, family life, social life, work life…
Appendix K: Letters to and from LREC

University of Leicester

Please reply to:
Sarah Crozier

email: [redacted]

Friday 14th December 2012

To Whom It May Concern:

Please find enclosed all supporting documentation for the below research project Research Ethical Committee application to attain ethical permission in order to proceed to R&D approval:

Title: Experiences of Spousal Caregivers of Individuals with Huntington's Disease through Pre and Post Clinical Diagnosis and the Effect of the Caregiving Process

Chief Investigator: Sarah Crozier

REC Reference No: 13/EM/0014

If there are any queries or questions regarding the application please feel free to contact me on the above email address for a prompt response. I look forward to hearing from you in due course.

Yours sincerely

Sarah Crozier
Trainee Clinical Psychologist
21 December 2012

Miss Sarah Crozier
Trainee Clinical Psychologist
Leicestershire Partnership Trust
104 Regent Road
Leicester
LE1 7LT

Dear Miss Crozier,

Study title: Experiences of Spousal Carers of Individuals with Huntington’s Disease through Pre and Post Clinical Diagnosis and the Effect of the Caregiving Process

REC reference: 13/EM/0014
IRAS project ID: 119814

Thank you for your application for ethical review, which was received on 17 December 2012. I can confirm that the application is valid and will be reviewed by the Committee at the meeting on 04 January 2013.

Meeting arrangements

The meeting will be held in [location] on 04 January 2013. The Committee would find it helpful if you could attend the meeting to respond to any questions from members. Other key investigators and a representative of the sponsor are also welcome to attend. This may avoid the need to request further information after the meeting and enable the Committee to make a decision on the application more quickly.

If you have a disability and need any practical support when attending the REC meeting you may wish to contact the REC office so appropriate arrangements can be made if necessary.

If you are unable to attend the meeting the Committee will review the application in your absence.

The review of the application has been scheduled for 11.30. Please note that it is difficult to be precise about the timing as it will depend on the progress of the meeting. We would kindly ask you to be prepared to wait beyond the allocated time if necessary.

Please let me know whether or not you would be available to attend the meeting or be available on the telephone.

Committee meetings are occasionally attended by observers, who will have no vested interest in the applications under review or take any part in discussion. All observers are required to sign a confidentiality agreement.
Participation in Ethics Officer Pilot

Please note this REC is taking part in the ethics officer pilot, to consider whether it is possible to increase favourable opinion rates, improve timelines and reduce the burden on coordinators and researchers alike by having an Ethics Officer, an experienced representative of the National Research Ethics Service (NRES) consider the application prior to review by the REC. As part of the pilot, [Name] may contact you or a member of the research team via phone or email to discuss your application. Please inform the coordinator if you are not the most appropriate person for the ethics officer to contact and provide details of a suitable alternate member of the research team or sponsor. Attached is a frequently asked questions guidance document for researchers and further information is available on the website (http://www.nres.nhs.uk/applications/hra-ethics-officer-pilot). If you require any further clarification, please contact [Name] HRA Board Secretary and CE Business Manager who will be happy to discuss the pilot with you further.

Documents received

The documents to be reviewed are as follows:

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<th>Date</th>
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<td>Evidence of insurance or indemnity</td>
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<td>Interview Schedules/Topic Guides</td>
<td>2</td>
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<td>Investigator CV</td>
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<td>Other: Service user reference group review feedback</td>
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<td>21 November 2012</td>
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<td>Other: Flowchart of protocol</td>
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<td>Participant Consent Form</td>
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<tr>
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<td>119814/394470/1/130</td>
<td>03 December 2012</td>
</tr>
<tr>
<td>Referees or other scientific critique report</td>
<td></td>
<td>08 November 2012</td>
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</table>

No changes may be made to the application before the meeting. If you envisage that changes might be required, we would advise you to withdraw the application and re-submit it.

Notification of the Committee’s decision

You will receive written notification of the outcome of the review within 10 working days of the meeting. The Committee will issue a final ethical opinion on the application within a maximum of 80 days from the date of receipt, excluding any time taken by you to respond fully to one request for further information or clarification after the meeting.

R&D approval
All researchers and local research collaborators who intend to participate in this study at sites in the National Health Service (NHS) or Health and Social Care (HSC) in Northern Ireland should apply to the R&D office for the relevant care organisation. A copy of the Site-Specific Information (SSI) Form should be included with the application for R&D approval. You should advise researchers and local collaborators accordingly.

The R&D approval process may take place at the same time as the ethical review. Final R&D approval will not be confirmed until after a favourable ethical opinion has been given by this Committee.

For guidance on applying for R&D approval, please contact the NHS R&D office at the lead site in the first instance. Further guidance resources for planning, setting up and conducting research in the NHS are listed at ftp://www.rdforum.nhs.uk

There is no requirement for separate Site-Specific Assessment as part of the ethical review of this research. The SSI Form should not be submitted to local RECs.

Communication with other bodies

All correspondence from the REC about the application will be copied to the research sponsor and to the R&D office for [redacted]. It will be your responsibility to ensure that other investigators, research collaborators and NHS care organisation(s) involved in the study are kept informed of the progress of the review, as necessary.

We are pleased to welcome researchers and R & D staff at our NRES committee members' training days – see details at http://www.hra.nhs.uk/hra-training/

13/EM/0014 Please quote this number on all correspondence

Yours sincerely

[Signature]

Committee Co-ordinator

Email: [redacted]

Enclosure: Further information about REC membership and meeting arrangements

Copy to: Sponsor/R&D Contact - [redacted]
15 January 2013

Miss Sarah Crozier
Trainee Clinical Psychologist
Leicestershire Partnership Trust
104 Regent Road
Leicester
LE1 7LT

Dear Miss Crozier,

Study Title: Experiences of Spousal Carers of Individuals with Huntington's Disease through Pre and Post Clinical Diagnosis and the Effect of the Caregiving Process

REC reference: 12/EM/0014
IRAS project ID: 119814

The Research Ethics Committee reviewed the above application at the meeting held on 04 January 2013. Thank you for attending to discuss the application.

Documents reviewed

The documents reviewed at the meeting were:

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<tr>
<td></td>
<td>7017/130</td>
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</table>

A Research Ethics Committee established by the Health Research Authority
reason for excluding anyone who is not White British was not valid. You explained that
the university had recommended she only include White British people to avoid
instances seen in some dementia studies where religious beliefs of ethnic minority
groups had a big impact on the difference in care. The Committee pointed out that white
British people could also have religious beliefs that might influence their attitudes to
care. The Committee stated that being inclusive was quite important and that the study
sample was not homogenous where sex or sexual orientation was concerned. There
was no valid reason for exclusion of other ethnic groups. The Committee told you the
exclusion criteria should be non English speakers only. You said you were happy to
comply with this request.

The Committee is unable to give an ethical opinion on the basis of the information and
documentation received so far. Before confirming its opinion, the Committee requests that you
provide the further information set out below.

Authority to consider your response and to confirm the Committee’s final opinion has been
delegated to the Chair.

Further Information or clarification required

1. Please remove White British from the exclusion criteria and instead make it clear that the
study will only exclude non-English speakers.

2. Please change the word “spousal” to “partner” in the research title on all study
documents.

3. Please make the following changes/revisions to the Participant Information Sheet.
   a) Page numbers should be added to the document.
   b) A time limit for withdrawal after the interview should be explicitly stated. E.g. 3
days after the interview.
   c) On page 1 under section “Why have I been chosen?” wording should be added to
      make it clear that the PIS has been sent by someone within the HD service so
      that there is no suggestion the researcher has already had access to their
      personal details.
   d) It should be added that the NRES Research Ethics Committee East Midlands-
      Leicester has reviewed the study.
   e) It should be included that participants will be asked to complete HADS and
      Demographic questionnaires.
   f) It should be added that interviews taking place at home should be in a private
      place.

4. Please make the following changes/revisions to the Consent Form.
   a) The first three paragraphs should be removed and added to the Participant
      Information Sheet.
   b) A point about access to study documents by regulatory authorities including the
      NHS trust should be added.
   c) A point giving consent for the use of direct quotations should be added.
   d) The version and date of the PIS that has been read should be included in point 1.
Provisional opinion

- The Committee asked why you intended to exclude anyone who was not White British from participation within the study. You said the inclusion of only White British was in order to maintain a homogenous participant group.

- The Committee commented on the fact that the HADS and demographic questionnaire were not mentioned within the submission and asked what you would do if someone came back as clinically depressed. You explained that anyone displaying signs of depression would be referred back to their GP or service.

- The Committee asked you if generational involvement mentioned within the Participant Information Sheet is a link to genetics. You confirmed that this was the case.

- The Committee asked about the peer review comments on use of male and female in relation to a relatively homogenous group. You said you did look at this but agreed that in the case of such a rare disease it would be better to include both male and females.

- The Committee told you that the word 'spousal' did not apply to same sex couples and with the hopes of not excluding this group it ought to be changed to 'partner'. You said you would change this.

- The Committee commented on the offer of withdrawal anytime after the interview and said this could cause loss of valuable data and instead suggested you set a time limit for this giving an example of 3 days after the interview. You said this had been your first thought and were happy to change this. The Committee went on to state that adding a limit would secure anonymity as, if you wanted it to be truly anonymous any links should be destroyed.

- The Committee asked about transcription and what level of data security you on your home computer. You told the Committee your computer is password protected and any transcribed data would be kept on an encrypted pen drive.

- The Committee stated that there was a possibility of arriving at a home for an interview and being asked by the carer if the participant could sit in as some carers would be anxious leaving the partner unattended and asked what you would do if this situation arose. You said you would probably suggest that the interview was rescheduled and hope to avoid interviewing in that setting but accepted that in such a situation you would have to be flexible.

- The Committee discussed minimal demographic data being sent out with study results and stated that this would have to be very minimal with a sample size of 4-6 participants. You accepted this.

- The Committee asked you to transfer the three statements of Information on the Consent Form to the Participant Information. You confirmed this would be done.

- The Committee went back to the ethnicity issue with the inclusion criteria and stated the
If you would find it helpful to discuss any of the matters raised above or seek further clarification from the Assistant Coordinator, you are welcome to contact [redacted] or via the email address below.

When submitting your response to the Committee, please send revised documentation where appropriate underlining or otherwise highlighting the changes you have made and giving revised version numbers and dates.

If the committee has asked for clarification or changes to any answers given in the application form, please do not submit a revised copy of the application form; these can be addressed in a covering letter to the REC.

The Committee will confirm the final ethical opinion within a maximum of 80 days from the date of initial receipt of the application, excluding the time taken by you to respond fully to the above points. A response should be submitted by no later than 14 February 2013.

Membership of the Committee

The members of the Committee who were present at the meeting are listed on the attached sheet.

A member of the Committee sits on the DCInPey board but has had no involvement in the design of the study. The committee agreed that it was fine for this member to remain in the discussion of the application.

Statement of compliance

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

13/EM/0014 Please quote this number on all correspondence

Yours sincerely

[Redacted]

Chair

Email: [Redacted]

Enclosures: List of names and professions of members who were present at the meeting and those who submitted written comments.

Copy to: [Redacted]
NRES Committee East Midlands - Leicester
Attendance at Committee meeting on 04 January 2013

## Committee Members:

<table>
<thead>
<tr>
<th>Name</th>
<th>Position</th>
<th>Present</th>
<th>Action</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Radiation Protection Advisor and Senior Lecturer (retired)</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Research Coordinator</td>
<td>Yes</td>
<td></td>
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<tr>
<td></td>
<td>Senior Lecturer</td>
<td>Yes</td>
<td></td>
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<tr>
<td></td>
<td>Implementation Fellow</td>
<td>Yes</td>
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<tr>
<td></td>
<td>Medical Statistician</td>
<td>Yes</td>
<td></td>
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<tr>
<td></td>
<td>Principal Lecturer in Clinical Pharmacy &amp; Pharmacy Practice</td>
<td>Yes</td>
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<tr>
<td></td>
<td>Reverend</td>
<td>Yes</td>
<td></td>
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<tr>
<td></td>
<td>Education and Practice Development Lead for Acute Care</td>
<td>Yes</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Senior Lecturer in Psychology</td>
<td>Yes</td>
<td></td>
</tr>
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</table>

## Also in attendance:

<table>
<thead>
<tr>
<th>Name</th>
<th>Position (or reason for attendance)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Assistant Committee Co-ordinator</td>
</tr>
<tr>
<td></td>
<td>Ethics Officer</td>
</tr>
</tbody>
</table>
University of Leicester

Please reply to:
Sarah Crozier

email:
Friday 18th January 2013

To Whom It May Concern:

Please find enclosed amended supporting documentation for the below research project Research Ethical Committee application to obtain ethical permission in order to proceed to R&D approval including protocol, participant information sheet and participant consent form:

Title: Experiences of Partner Caregivers of Individuals with Huntington’s Disease through Pre and Post Clinical Diagnosis and the Effect of the Caregiving Process

Chief Investigator: Sarah Crozier

REC Reference No: 13/EM/0014

Changes of note include the change of ‘spousal’ to ‘partner’ carers and the removal of White British participants in the inclusion criteria in order to be more inclusive.

If there are any queries or questions regarding the application please feel free to contact me on the above email address for a prompt response. I look forward to hearing from you in due course.

Yours sincerely

Sarah Crozier
Trainee Clinical Psychologist
25 January 2013

Miss Sarah Crozier
Trainee Clinical Psychologist
Leicestershire Partnership Trust
104 Regent Road
Leicester
LE1 7LT

Dear Miss Crozier,

Study title: Experiences of Spousal Carers of Individuals with Huntington's Disease through Pre and Post Clinical Diagnosis and the Effect of the Caregiving Process

REC reference: 13/EM/0014
IRAS project ID: 119814

Thank you for your letter of 18 January 2013, responding to the Committee's request for further information on the above research and submitting revised documentation.

The further information has been considered on behalf of the Committee by the Chair.

We plan to publish your research summary wording for the above study on the NRES website, together with your contact details, unless you expressly withhold permission to do so. Publication will be no earlier than three months from the date of this favourable opinion letter. Should you wish to provide a substitute contact point, require further information, or wish to withhold permission to publish, please contact the Co-ordinator [redacted].

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.

Ethical review of research sites

NHS 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.

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

Management permission ("R&D approval") should be sought from all NHS organisations involved in the study 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.cfsforum.nhs.uk.

Where a NHS organisation's role in the study is limited to identifying and referring potential participants to research sites ("participant identification centre"), guidance should be sought from the R&D office on the information it requires to give permission for this activity.

For non-NHS sites, site management permission should be obtained in accordance with the procedures of the relevant host organisation.

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

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:

<table>
<thead>
<tr>
<th>Document</th>
<th>Version</th>
<th>Date</th>
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</thead>
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<td>Covering Letter</td>
<td></td>
<td>14 December 2012</td>
</tr>
<tr>
<td>Evidence of Insurance or Indemnity</td>
<td></td>
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<tr>
<td>Interview Schedule/Topic Guides</td>
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<td>16 November 2012</td>
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<td>Investigator CV</td>
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<tr>
<td>Letter from Sponsor</td>
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<td>06 December 2012</td>
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<tr>
<td>Other: CV - Dr Robertson</td>
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<tr>
<td>Other: Service user reference group review feedback</td>
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<td>21 November 2012</td>
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<tr>
<td>Other: Flowchart of protocol</td>
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<td>29 November 2012</td>
</tr>
<tr>
<td>Participant Consent Form</td>
<td>3</td>
<td>18 January 2013</td>
</tr>
<tr>
<td>Participant Information Sheet</td>
<td>3</td>
<td>18 January 2013</td>
</tr>
<tr>
<td>Protocol</td>
<td>6</td>
<td>18 January 2013</td>
</tr>
<tr>
<td>Questionnaire: HADS</td>
<td></td>
<td></td>
</tr>
<tr>
<td>REC application</td>
<td>118814/5884701/1/130</td>
<td>03 December 2012</td>
</tr>
<tr>
<td>Referee or other scientific critique report</td>
<td></td>
<td>06 November 2012</td>
</tr>
<tr>
<td>Response to Request for Further information</td>
<td></td>
<td>18 January 2013</td>
</tr>
</tbody>
</table>
Statement of compliance

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

After ethical review

Reporting requirements

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
- Notification of serious breaches of the protocol
- 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.

Feedback

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.

Further information is available at National Research Ethics Service website > After Review
We are pleased to welcome researchers and R & D staff at our NRES committee members' training days – see details at http://www.hra.nhs.uk/hra-training/

With the Committee's best wishes for the success of this project.

Yours sincerely

[Signature]

Chair

Email:

Enclosures: *After ethical review – guidance for researchers*

Copy to: Sponsor/R&D Contact -
Appendix L: IPA Strategy Process

The common processes and principles described by Smith et al. (2009) were followed during the process of IPA analysis adhering to the six main steps of: reading and re-reading; initial coding; developing emerging themes; searching for connections across emerging themes; moving to the next case; and looking for patterns across cases.

Initial reading and re-reading of individual transcripts was completed before conducting a close, line-by-line analysis producing phenomenological coding of the individual’s experience (Larkin et al., 2006), focused on making initial descriptive and linguistic comments. The second stage of analysis involved the identification of patterns within the initial phenomenological codes for each individual transcript (Eatough & Smith, 2008). The process of combining the phenomenological coding with the researchers psychological knowledge allowed for the development of interpretative codes giving meaning to the respondents’ experiences described (Larkin et al., 2006; Smith, 2004).

This two-stage interpretative process (Smith & Osborn, 2008) was completed for each individual transcript to stay true to ideographic nature of IPA and prevent the unique elements of individual respondents’ experiences from being lost. After its completion, patterns and themes were compared across respondents for recognition of commonalities and
nuance which contributed to the development of themes and the narrative of the relationships between them. Through this process five superordinate themes were identified across the six respondents’ experiences with 18 sub-themes across these super-ordinate themes which helped explain the idiosyncrasies found between respondents.

Throughout the whole research process the researcher kept a reflective journal of their perceptions, conceptions and processes as suggested by Smith (2007), which contributed to the critical appraisal of this thesis.

References


Appendix M: Example of Initial Coding of Transcript

yesterday and he didn’t even notice and you think you know...one of these sort of things why bother sort of things.

Is that something that’s been a change in your relationship?

Umm...I guess it is because at one time I used to buy loads and loads of clothes and he used to say have you bought clothes again. And I’d say no I’ve had these for ages you know that sort of thing and he used to say it was the joke that I’ve got clothes for every occasion sort of thing. And he doesn’t even notice now what I’m wearing or if...he does when I’ve been to have my hair done he’ll come back...when I come back he’ll say your hair looks nice but it’s because he’s been here on his own, he’ll have known I’d been to the hairdressers you know but...I guess that is a change. It’s quite a big change because at one time he was always sort of...you’d get compliments from him and you’d get you know...like I said we always used to talk about everything but now you just can’t talk about anything. You know so there’s been a big change there.

Are there any other changes you’ve noticed, changes in your life or in [name]?

Umm...only the fact that he’s got absolutely no interest in anything. You know whereas before he would constantly be planning and doing something. I mean we’ve had...I think this has been our sixth or seventh house and...even we’ve had a brand new house that we were the only people to have lived
Appendix N: Researcher Reflexivity Statement

The researcher has knowledge and experience in a clinical context of working with individuals diagnosed with HD and their families. The researcher also has personal experience of caring for a family member with a neurodegenerative condition (vascular dementia). It is this personal experience that motivated the researcher to explore firstly the literature basis around dementia and subsequently the limited research base regarding HD for which dementia research was heavily relied upon before conducting this research.

The researcher recognised that these experiences and knowledge will have influenced and shaped the current research findings with the pre-existing assumptions such as:

- Degenerative and genetic conditions affect the whole family;
- Caring for a relative with a degenerative condition can be difficult for most if not all individuals; and
- Availability of services designed with carer needs, alongside those of the individuals diagnosed with the condition, would benefit carers...
Appendix O: Full Chronology of Research Process

Jan 2012 - May 2012
- Consultation with research supervisor
- Initial research protocol

May 2012
- Developing research protocol

Nov 2012
- Internal peer review at the University of Leicester
- Service User Reference Group (SURG) review
- Preparation of application for LREC

Dec 2012
- LREC application
- R&D application

Jan 2013
- LREC meeting 4th January 2013, favourable opinion received 25th January 2013
- R&D approval received 28th January 2013/1st February 2013

April 2013 - Nov 2013
- Recruitment and interviewing participants
- Transcription

Jan 2014 - March 2014
- Analysis

Nov 2013 - April 2014
- Write up period

April 2014
- Submission of thesis to University of Leicester

May 2014 - July 2014
- Viva preparation

July 2014 - Sept 2014
- Dissemination of findings
- Preparation for poster presentation and publication paper
## Appendix P: Frequency of Themes Across Respondent's Transcripts

<table>
<thead>
<tr>
<th>Themes</th>
<th>Transcript No.</th>
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<td>3</td>
<td>4</td>
<td>5</td>
<td>6</td>
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<tr>
<td><strong>A. Collective vs. Individual Care</strong></td>
<td>X</td>
<td>X</td>
<td>X</td>
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<tr>
<td>A.1 Dichotomous roles</td>
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<td>X</td>
<td>X</td>
<td>X</td>
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<td>X</td>
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<tr>
<td>A.2 Protection</td>
<td>X</td>
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<td>A.3 Isolation of the HD family</td>
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<td><strong>B. The invisible partner</strong></td>
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<td>B.1 Not noticed within the relationship</td>
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<tr>
<td>B.2 Not noticed outside of the relationship</td>
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<td>B.3 Gender identification</td>
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<td>B.4 Self-sacrifice</td>
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<td><strong>C. The emotional roundabout</strong></td>
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<td>C.1 Impact of diagnosis: Shock</td>
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<td>C.2 Hopelessness and wishful thinking</td>
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<td>C.3 Circular emotions</td>
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<td>D.1 Loss of life as it was</td>
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<td>E.5 Part of something bigger</td>
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