Resignation, in the absence of cure: A narrative study on the illness experience of Huntington's Disease.

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PRESENTATIONS AND PUBLICATIONS RELEVANT TO THE STUDY

PRESENTATIONS:

Carreon, R.P. (2022) I am not Huntington's: reflections on person-centred care and physiotherapy practice, Physiotherapy Working Group, European Huntington Disease Network Annual General Meeting (Oral Presentation)

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Carreon, R.P. (2020) Narratives of Huntington's Disease: from concept to analysis. Faculty of Health Doctorate Conference, Liverpool John Moores University (Oral presentation)

Carreon, R.P. (2020) Is it over the phone? Studying illness narratives in the midst of Covid-19. Northwest England PhD qualitative online research group, Edge Hill University (Oral presentation)

Carreon, R.P. (2020) A question on validity: remote data collection methods. Northwest England PhD qualitative online research group, Edge Hill University (Oral presentation)

Carreon, R.P. (2020) Three-minute thesis. A flip of a coin: Living with Huntington's disease. Liverpool John Moores University (Oral presentation) (Faculty of Health Winner)

Carreon, R.P. (2020) Every story is us: the narrative case series of Huntington's Disease. Liverpool Research Café: communicating research. Liverpool John Moores University (Oral presentation) (Recipient of LJMU Post Graduate Researcher Outstanding Communicator Award 2020)

Carreon, R.P. (2020) Huntington's disease: from the lens of a physiotherapist and PhD researcher. St. Andrew's Health Care, Northampton, Physiotherapy and Allied Health. (Oral presentation)

Carreon, R.P. (2020). Systematised literature review: an example from a Huntington's disease study. Liverpool John Moores University. (Oral presentation)

Carreon, R.P. (2020) Conducting remote interviews at the time of pandemic: ethical and practical considerations. Liverpool John Moores University (Oral presentation)

Carreon, R.P. (2019) Huntington's disease and neurological conditions: the need for public health awareness. Public Health Institute PhD Symposium, Liverpool John Moores University (Oral presentation)

Carreon, R.P. (2019) The illness experience of Huntington's disease: findings from a literature review. Public Health Institute PhD Symposium, Liverpool John Moores University (Poster)

Carreon, R.P. (2019) Living under the shadow of Huntington's disease. Research and Innovation Conference, Liverpool John Moores University. (Scholarly snap) (People's choice winner and highly commended award)

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CONFERENCE PUBLICATIONS:

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ABSTRACT

The illness experience of Huntington's Disease (HD), a rare genetic progressive fatal neurological disorder, receives scant attention in qualitative research despite the historical stigma and shame associated with the movement disorder, serious mental illness, and the pervasive suffering it causes to the individual and their families. This in-depth narrative study aims to give voice to the person with HD and explore how they construct their illness narratives. It draws upon illness storylines and dialogic approaches to narrative analysis. A scoping literature review on HD illness experience and a Participatory Research Exercise (PRE) with HD stakeholders informed the creation of the narrative cases. A total of nineteen participants were involved at various stages of the study: ten participants in the PRE, and nine in the narrative cases.

At the heart of the study were three stories of people with HD, supplemented by their family caregivers and health care professionals. Multiple sources and methods of data collection were used, including biographical interviewing, genograms, life story booklets and other personalised artefacts. The tailored narrative methods supported the narration and construction of the story of the person with HD sustained during 18 months of participation in the study.

The HD stories revealed the unique illness experience of three people in different disease trajectories. The interviews were analysed using Arthur Frank's (1995) archetypal storylines and an additional storyline was added due to the unique nature of the HD experience. The study proposes the novel storyline of Resignation, for Restitution is no longer possible for this terminal illness, and has receded into the background of participants' stories.

The study highlights how the narrative methodology can provide valuable insight into the HD illness experience and contributes to a limited HD qualitative literature on the use of multiple data sources through a longitudinal approach in studying illness experience. The study also begins to fill the gap in physiotherapy knowledge on the illness experience of HD. From a physiotherapy perspective, listening to storylines can provide a rich understanding of people's experiences by examining social, cultural and contextual factors which can help clinicians develop educational resources to inform better patient care and help support advocacy efforts.

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LIST OF ABBREVIATIONS

AD	Alzheimer's Dementia
ADL	Activities of Daily Living
CAG	Cytosine, Adenine, and Guanine (three DNA building blocks)
COPD	Chronic Obstructive Pulmonary Disease
Covid-19	Severe acute respiratory syndrome Coronavirus 2
DNA	Deoxyribonucleic acid, a chemical that contains genetic information
EHDN	European Huntington's Disease Network
GP	General Practitioner
HCA	Health Care Assistant
HCP	Health Care Professional
HD	Huntington's Disease
HDA	Huntington's Disease Association
HRQOL	Health Related Quality of Life
HTT	Huntingtin Protein
HIV	Human Immunodeficiency Virus
LOK	Lasting Power of Attorney
MCA	Mental Capacity Assessment
MDT	Multi-Disciplinary Team
MHA	Mental Health Act
MND	Motor Neuron Disease
MS	Multiple Sclerosis
NICE	National Institute of Clinical Excellence
NOK	Next of Kin
NC	Narrative Case
OT	Occupational Therapist
PD	Parkinson's Disease
PEG	Percutaneous Endoscopic Gastrostomy
PLTNC	Progressive Long Term Neurological Conditions
PT	Physiotherapist
QOL	Quality of life
RCT	Randomised Controlled Trial
SALT	Speech and Language Therapist
SCI	Spinal Cord Injury
TFC	Total Functional Scale
UHDRS	Unified Huntington's Disease Rating Scale

DEDICATION

To Jen, Martin, and Daniel.

In the end, we're all stories.

CHAPTER 1: INTRODUCTION

What we want to know or seek to explore begins with a question. And a question begins with a 'quest' (being literal). The quest of this thesis is to better understand what it is like living with Huntington's Disease (HD) and how people with HD construct their illness narratives.

Huntington's Disease or HD is a rare genetically caused brain disorder that has no cure. Research indicates that families and caregivers face difficulty in talking about HD due to the limited understanding of the illness shared by the public and some health professionals (Skirton et al., 2010; Parekh, Praetorius and Nordberg, 2017). When asked "what is HD?", I often find it easier to compare it with more common neurological conditions. HD combines the features of Parkinson's Disease as a movement disorder, the cognitive deficits of Alzheimer's Dementia, and the psychiatric issues associated with Schizophrenia. I would also add that HD is a debilitating illness or a perfect storm; gradually the symptoms appear, almost unnoticeable and subtly progress over time, eventually leaving the hapless sufferer completely incapacitated- unable to eat, walk and talk. They become almost unrecognisable from their former self. But what makes HD different from the other brain diseases is that HD is purely genetic, and it can strike at any age. HD has clinical and public health implications (Bird, 2019). It is a model brain disease for it merges key neurological symptoms, disrupting the connection between movement, behaviour and thinking. Equally, the wider social consequences allow us to better understand how individuals, families and communities deal with a hereditary stigmatising mental illness. It is my view that no other disease bridges the disciplines of genetics, neurology, psychiatry, and sociology more than HD.

HD has received much interest in scientific research as a disorder governed by genetics, and literature predominantly focuses on the disease biomedical aspect. The advances of medicine and technology have significantly improved our clinical knowledge, however there is very limited research on the HD subjective experience (Zarotti et al., 2022), especially in the advanced stages of disease progression. Hence, this thesis is a quest in its own right, asking how we can learn more about HD as an 'illness' rather than a 'disease'? (Kleinman, 1988; Sacks, 2014). I turn to Arthur Frank's (2020) advice that in approaching a question of 'a significant clinical, scholarly, and personal ideal is thinking with a story' (p.1). This led to the fundamental quest of this thesis which was to present HD stories using the voices of those who live with this incurable illness.

PURPOSE OF THE STUDY

This thesis builds on some of the work described in qualitative literature on illness experience and fills in gaps in HD literature. Moreover, there is an imperative need to better understand the subjective experience of HD in order for clinicians to better provide holistic care for patients and their families. There is a dearth of in-depth qualitative research in HD considering the complex issues around the disease and the long progression of the illness which can affect generations of families. Narrative research on the illness experiences provides both personal and social context but, with the exception of one study

(Schwartz, 2010) which explored a narrative approach to HD diagnosis, it is currently lacking in this area in comparison to other neurological conditions (Audulv, Packer and Versnel, 2014). This thesis endeavours to contribute to the body of literature on illness experience (Thorne et al., 2002; Soundy et al., 2013) and the proposals made by Audluv et al (2014) and Zarroti et al (2022) that more qualitative methods are needed to explore the subjective experience of living with HD.

The purpose of this study is to explore the illness experience of people with HD in different manifest stages. There is an empirical need to explore HD experiences beyond diagnosis and to understand how individuals narratively construct their illness experience. In traditional research, researchers look for answers to particular questions using different forms of data collection (Kramp, 2004), but what makes narrative researchers different is that they construct data in what 'we call stories' (p.112). I set out to study stories of HD participants, record our interactions through fieldnotes, make use of available documents and artefacts generated through interviews and other research tools. This multi-modal approach speaks to the preference of the participants, availability of resources and practicability of use.

The study was guided by a narrative approach underpinned by Arthur Frank's theoretical framework (Frank, 1995; Frank, 2010). In applying Frank's lens in HD illness experience, I aim to expand our existing knowledge and offer a specific contribution to HD qualitative literature. Based on my knowledge, this is the first HD narrative study to apply Arthur Frank's (1995, 2010) methodology in studying the illness experience of HD. At the heart of this study is the story of the person with HD, using multiple methods and multiple sources of data sustained over 18 months. The unique emphasis of this study is the collaborative and participatory aspect, in which the participants help shaped the telling of their story, including the choice of methods and how these were then adapted in the course of the research. This approach generated a rich contextual understanding of the HD illness experience in various trajectories.

THE RESEARCHER'S CONTEXT

'If you want to know me then you must know my story, for my story defines who I am. And if I want to know myself, to gain insight into the meaning of my own life, then I, too, must come to know my own story'. (McAdams 1993, p.11)

My interest in HD started as a clinician, a specialist physiotherapist working with neurological conditions. I have studied HD mainly as a genetic movement disorder (Bates et al., 2015). I was qualified in 2003 and it wasn't until 2010 that I met my first HD patient. A UK survey (Ho, 2018) pointed out that only 12 percent of HD patients have accessed physiotherapy services, hence it is uncommon to see an HD patient in clinical practice. I clearly remembered how I felt when I met my first patient with HD: a sense of curiosity and trepidation at treating a rare condition. I observed that HD characteristics and care profiles significantly differ from other progressive long term neurological conditions (PLTNC). I initially had difficulty engaging HD patients with structured physiotherapy exercises, mainly due to their behavioural symptoms, which led me to gain further training on mental health and therapeutic behavioural interventions. The mental health

courses outside of my usual physiotherapy practice helped me gain confidence and develop strategies to motivate patients with exercise, counteract apathy and understand various behavioural issues associated with HD. As I began my career specialising in complex neurological care, the service grew, with an increasing number of HD referrals. I observed within my workplace that caring for patients with HD has its own set of challenges, due to their multiple complex needs that consequently require 'multifaceted care which can be complex to deliver' (Mestre and Shannon 2017, p.114), more so around the behavioural and psychological aspects of the illness (Craufurd and Snowden, 2002; Ghosh and Tabrizi, 2018; Zarotti et al., 2020). I felt that I needed specific knowledge about HD, and I reached out to HD organisations such as the European HD Network (EHDN) and Huntington's Disease Association (HDA) and found their resources very valuable. I wasn't surprised that studies of people with HD and their families demonstrated a lack of knowledge among health professionals (Domaradzki, 2015; Anderson et al., 2019), which I also observed early on in my career.

As I addressed my professional development, I completed a Post Graduate Certificate in Clinical Examination to hone my assessment skills before I embarked on my Master's (MSc) in Public Health. During my MSc dissertation I explored the needs of HD professional caregivers and discovered the nurses and health care assistants (HCAs) perceived barriers and facilitators of providing care. I chose nurses and HCAs because their working shifts meant they are with HD patients for 12 hours in comparison to an average therapy time of an hour per patient. What surprised me about the study was that it was not necessarily their training nor education that they valued, but how much they learned from HD patients. However, their answers were generally vague and varied on how they viewed learning from their patients. This spurred both my interest in qualitative research and studying HD. Hence, this PhD study stemmed from the main findings of the MSc research that the 'HD patient is the biggest teacher' (Carreon et al., 2018, p.76).

I recalled how the case histories and stories of my HD patients personally intrigued me. There was never a straightforward trajectory of how they discovered the HD gene in the family, how their symptoms affected their jobs, how their illness fragmented their personal relationships nor how they ended up in institutionalised care. Despite being drawn to their and their caregivers' stories, my clinical training impelled me to focus on the pathophysiological care and treatment. But whenever I met a new HD patient, they brought their remarkable illness journey. I have assessed HD patients in prisons, psychiatric units and in older people's homes. Their social and educational backgrounds widely differ, as well as their ages with some as young as 21. I had heard that, 'if you've seen one HD patient, you have seen just one patient' and there is truth to this as everyone is unique and HD symptoms are so diverse. Yet, there was resonance in their stories of living with an illness that is misunderstood (Bird, 2019), and examples include the repeated accounts of being accused of drunkenness, dysfunctional families, and their unknown histories (Wexler, 2012; Bird, 2019). Embedded within their stories were HD secrecy and stigma that has been a theme in popular culture (Wexler, 2014) and in HD literature (Wexler, 2010; Williams et al., 2010; Wauters and Van Hoyweghen, 2021). While the erratic movements can be often their source of anxiety and ostracism (Bachoud-Lévi et al., 2019), I noted that my patients' personalities transcended motor impairments or

occasional absences of speech and identified with a neurologist who once remarked 'there's something special about HD'. I also recalled a consultant to whom I spoke at an HD conference saying, 'Once you meet an HD patient, you'll never forget them'.

Indeed, HD has personally and professionally spurred my interest, beyond what George Huntington (1872) initially described as a medical curiosity. Like the clinicians Kleinman (1998) and Sacks (2014), I wanted to learn about HD more as an illness, than just a 'disease'. Illness, as distinguished by Kleinman (1998), describes how the affected person, family members or wider social system 'perceive, live with, and respond to symptoms and disability; the innately human experience of symptoms and suffering' (p.3). After I completed my MSc research and presented the findings at the EHDN Conference in Vienna, Austria, I explored with my supervisors PhD opportunities leading, in turn, to a grant from Liverpool John Moore's University Vice Chancellor Scholarship that made this study possible.

REFLEXIVITY

'No research is free of the biases, assumptions and personality of the researcher, we cannot separate self from those activities in which we are intimately involved'. (Sword 1999, p.277)

I commenced the study bringing with me my experiences, standpoints, and personal agendas. Finlay (2003) stated the 'the researcher is a central figure' (p.5) from the beginning to end of the study. What we bring to the study and our epistemologies influence what we see and define the methods we choose (Charmaz, 2015). Here I attend to Pillow's (2003) advice to use reflexivity to critically interrogate my research practice. However, Finlay (2003) noted the diverse theoretical positions and definitions inform how different researchers practice reflexivity. Therefore, in this study I adopt an iterative reflexive practice to examine my own subjectivity and how this affects and shapes the study processes and outcomes. In addition, I will use Finlay's (2003) notion of 'reflexivity as an inter-subjective reflection' (p.8) to examine the research relationship between myself, the participants, and the research process. This approach provides a study focus that employs 'collaborative inquiry', as I 'look inward for personal meanings and outward into the realm of shared meanings, interaction, and discourse' (Finlay and Gough 2008, p.10).

In this thesis, reflexivity informs a broader discussion of the epistemological and axiological components of research knowledge (Bergner 2015). I locate my personal experience to establish context which readers can engage with the 'evolving search for new meaning and creation of a final research product' (Sword 1990, p.270). By reflexively sharing my own experiences in this study, I hope to explore how my own subjectivities might have influenced the research process and informed the production of knowledge. With all these reasons, I write in first person taking account my own feelings and perspectives and reflect upon how my relationships with the participants and the research has evolved during the study.

OVERVIEW OF THE THESIS

My thesis is divided into seven chapters. Chapters 1 to 4 set the scene for the study. Chapter 1 takes the form of a brief introduction and outlines the thesis chapters. Chapter 2 contains the literature review, exploring extant HD literature and identifying gaps in our knowledge. The chapter begins with the perspectives of clinicians, how HD is historically viewed in medical literature and narrates its contemporary position in relation to current health policy and guidance. The literature review then focuses on the personal experiences of HD, largely drawn from the perspectives of family caregivers and this is followed by a scoping review on the illness experiences of individuals diagnosed with HD. My review of HD literature highlights areas in need of further research. These gaps informed the rationale of conducting this thesis, the methodology used and the development of the research question, which will be presented at the end of the literature review. Chapter 3 defines the general methodological approach to the study. It examines theoretical approaches in narrative research and illness narratives. This chapter begins with my epistemological and ontological perspectives and concludes with the development of the research question. Chapter 4 describes the inception of the narrative case design which employed multiple sources and methods of data collection. This chapter provides the details of study, the process of approvals, and the study's analytical approach.

Chapter 5 presents the study findings illustrated in stories. This is the core of the thesis describing a longitudinal study of the illness experience of three people with HD, alongside that of their families and of health professionals. The study applied Arthur Frank's theory of illness storylines and a dialogical approach to narrative analysis. The storylines were used as a heuristic device to provide structure and explore how people with HD construct their illness narratives.

Chapter 6 contains the discussion of the study findings and an explanation of how the stories relate to theory and current literature. In this chapter, the study will be assessed using relevant guidelines and to determine its strengths and limitations. The study's general and clinical implications will also be considered. Chapter 7 is the final chapter and will examine key issues in the thesis including reflexivity and ethics. The chapter also presents the study-specific contribution to literature, possible future directions, and my closing thoughts.

Overall, the narrative approach used in this study deepens our understanding of the HD illness experience from a variety of different perspectives. Although this thesis deviates from the traditional structure found in standard research texts, I hope to give an account of the research journey in an accessible format, and to describe how each undertaking has informed, adapted, and changed the research process over the course of time. This is my commitment to a narrative approach which I believe is an integral part of the research process, and which is well suited to the study of illness stories.

Frank asserts that illness is the loss of the destination and map, the purpose of stories is to repair the damage that illness has done to the ill person's sense of where she is in life, and where she may be going.

‘Stories are a way of drawing new ways and finding new destinations’ (Frank 1995, p. 53). And if you the reader of this thesis, seeks a new map to understand the illness experience of Huntington’s Disease, then come walk with me.

CHAPTER 2: LITERATURE REVIEW

CHAPTER OVERVIEW

To facilitate an understanding of the HD illness experience, a conceptual framework was developed to help navigate the literature (Figure 1). The literature review is divided into three general domains, these include 1) the clinical perspective highlighting the elements of HD history, disease pathology, contemporary treatment, and care, 2) the families' and caregivers' perspective exploring relational structures and experiences of caring for individuals with HD, and 3) the personal perspectives of people with HD, exploring available literature of the HD illness experience. It is hoped that, taken together, these multiple perspectives will enable an understanding of HD experiences and provide a means of identifying gaps in our current knowledge. The literature review will also set the scene for the next chapter, which explores the narrative approaches that constitute the study methodology of this thesis, and which touches upon illness narrative literature.

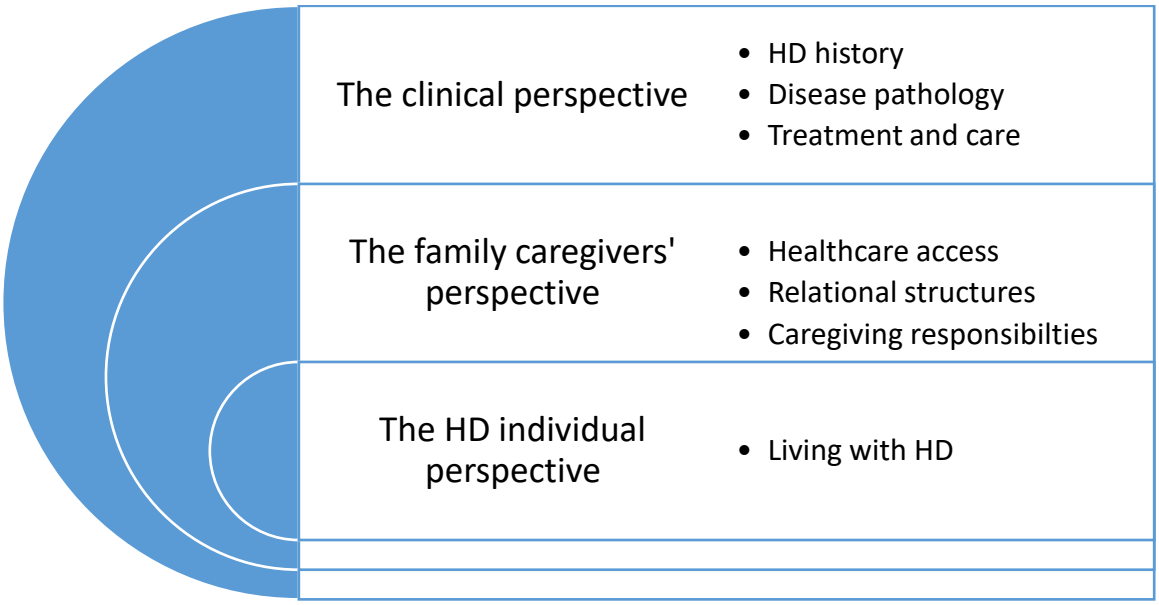


Figure 1 The three sides of HD: perspectives from clinicians, families and caregivers, and the HD individual.

THE SEARCH STRATEGY

The search strategy was developed from key search terms in each domain, and these were consequently adopted for each database. The four largest medical and nursing databases were searched: SCOPUS, Medline, CINAHL and Psych Info. Guidance on the search strategy was obtained from the academic liaison librarian in accessing relevant databases and search tools. Limits were placed on searches which included "full text", "English language" and date limit of 10 years from "2008 to current year." The majority of the

literature search was performed from October to December 2018, with further searches in March to May 2020, and updated regularly during the remainder of the PhD candidature.

Search strategies to identify relevant literature were developed using the SPIDER (Sample, Phenomenon of Interest, Design, Evaluation, Research type) Tool. The SPIDER strategy was developed to advance the PICO (Population, Intervention, Comparison, and Outcome) model for evidence-based medicine (Table 1). SPIDER is best suited for searching qualitative driven and mixed-method research questions (Cooke, Smith and Booth, 2012). The search terms used are listed in each relevant literature domain.

Table 1. **The construction and justification of the SPIDER search tool (Cooke, Smith and Booth, 2012).**

SPIDER	JUSTIFICATION
S- Sample	Smaller groups of participants tend to be used in qualitative research than quantitative research, so this term was deemed more appropriate.
PI- Phenomenon of Interest	Qualitative research to understand the how and why of certain behaviours, decisions, and individual experiences. Therefore, an intervention or exposure per se is not always evident in qualitative research questions.
D-Design	The theoretical framework used in qualitative research will determine the method that is used. As inferential statistics are not used in qualitative research, details of the study design will help to make the decisions about the robustness of the study and analysis. In addition, this might increase the detection of qualitative studies in the databases in which titles and abstracts are unstructured.
E- Evaluation	Qualitative research has the same end result as quantitative research methods: outcome measures. These differ depending on the research question and might contain more unobservable and subjective constructs when compared to quantitative research.
R- Research type	Three research types could be searched for: qualitative, quantitative, and mixed methods.

THE CLINICAL PERSPECTIVE

The SPIDER approach was used to identify the literature on the clinical aspect of HD. Key terms for Sample (S) “Huntington’s Disease” OR “HD”, Evaluation (E) key words included “pathology”, “symptoms”, “epidemiology”, “disease management” and “treatment”. Research type (R) include “quantitative”, “qualitative” and “mixed methods”. Phenomenon of Interest (PI) and Design (D) were not used as the search focus on observational studies. Limits were placed which included full texts only and articles published in the English language.

HD: LEARNING FROM HISTORY

Although early descriptions of HD already existed since at least the seventeenth century, it was the description of a 21 year old physician George Huntington in 1872, a disorder he referred to as a 'medical curiosity' that spurred the interest of the medical and scientific community (Lanska, 2000; Wexler, Wild and Tabrizi, 2016). In his paper "On Chorea" (Huntington 1872, reprinted in 2003 p. 111), Huntington noted that,

'There are three marked peculiarities in this disease: 1) Its hereditary nature. 2) A tendency to insanity and suicide. 3) Manifesting itself as a grave disease only in adult life... I have never known a recovery or amelioration of symptoms in this form of chorea; once begins it clings to the bitter end. No treatment seems to be of any avail, and indeed nowadays its end is so well known to the sufferer and his friends that medical advice is seldom sought. It seems at least to be one of the incurables.'

Huntington came from a family of physicians and he initially used observations and case history notes made by his father and grandfather (Durbach and Hayden, 1993). His longitudinal study of families in East Hampton, New York revealed the hereditary traits of the disease (Wexler, Wild and Tabrizi, 2016). Although Huntington at the time only acknowledged the adult onset, the disease also manifested itself, albeit rarely (around 10 percent of cases) in children termed as Juvenile HD (Quarrell, 2008). Huntington's comprehensive observations later earned his eponym (Walker, 2007) his portrait of the disease remains relevant 150 years later (Durbach and Hayden, 1993; Nance, 2017). Researchers argue that although Huntington's seminal work (1872) was not the first clinical account of a hereditary chorea (Lanska, 2000; Walker, 2007; Roos, 2010), it was the complete social, cultural, and medical account that defines the entity of the disease. The disease became known as 'Huntington's chorea', later changed in 1960 to 'Huntington's disease' (Wexler, Wild and Tabrizi, 2016).

Huntington's Disease, abbreviated as HD, is an incurable disease that is dominantly inherited, with progressive motor, cognitive and neuropsychiatric disorder affecting generations of afflicted families (Huntington, 1872; Bates et al., 2015). Huntington's original description of the disease included this element of inheritability which distinguished it from other neurological conditions (Harper, 1992; Harper, 2002). The hereditary characteristic has impelled a great deal of scientific work over the centuries which has led to the discovery of its causative gene (Nance, 2017). The significant events in the HD timeline are summarised in Table 2.

Table 2. Significant dates and events in HD timeline adopted from Walker (2007) and Harper (2002).

Year	Event
1374	Epidemic dancing mania detected.
1500	Paracelsus, a Renaissance alchemist (1493-1541), coined the term and suggested the brain origin of "Chorea" a dance like movement disorder.
1630s	"That disorder" and "Saint Vitus dance" were described by English colonists in Massachusetts, Connecticut, and New York.
1686	Thomas Syndeham, an English physician described post-infectious chorea.
1692	The Salem Witch Trials occurred in Salem, Massachusetts. It is believed that the "witches" are possessed by the devil, as they show erratic behaviours and chorea like-movements. Historians attributed these manifestations to HD.
1832	John Elliotson, an English physician identifies an inherited form of chorea.
1872	George Huntington, an American physician characterises HD.
1908	Mendelian dominant inheritance recognized.
1910-1911	Charles Davenport, an American eugenicist proposed compulsory sterilisation and immigration restriction for people with genetic diseases such as HD.
1933-1945	The German Reich (Nazi Germany) included HD in the race-hygiene policies. Historical accounts suggested that there were up to 3,500 who were compulsory sterilised. In 1941 alone, over a 100 people with HD were killed in one psychiatric unit.
1953	James Watson and Francis Crick, American scientists discovered the chemical structure of DNA.
1955	Americo Negrette, a Venezuelan physician described the unusually high number of cases of dancing mania, in Lake Maracaibo, Venezuela. The syndrome which locals call "El mal" (the bad), was later changed to an HD diagnosis.
1967	Woody Guthrie, the American poet and songwriter dies of HD. To promote public outreach, Guthrie´s wife, Marjorie, founded the Committee to Combat Huntington´s Disease (CCHD), now called the Huntington´s Disease Society of America (HDSA).
1972	The International Centennial Symposium on HD held the hundredth anniversary of George Huntington´s historic publication (1872).
1976	First animal model using Kainic acid was described.
1983	Localization of HD gene on chromosome 4 / discovery of gene marker.
1993	HD gene was isolated and identified.

	Clinical trials were formed by the Huntington Study Group.
1996	Development of a transgenic mouse model.
2000	Drugs screened for effectiveness in transgenic animal models.
2015	Beginning of HD human trials.
2017	First human trial of HD drug demonstrated that it reduces HD protein in the nervous system. Pope Francis met with a group of HD patients and families from around the world, a pharmaceutical company, Roche sponsored the documentary film: 'Dancing at the Vatican'.
2019	Roche and Ionis: GENERATION HD1- Worldwide trials begin.
2020	Covid-19 Pandemic hit Globally.
2021	GENERATION HD1 Drug trials halted.

A few of the many works by numerous researchers have deepened our knowledge of HD (Walker, 2007; Bates et al., 2015; McColgan and Tabrizi, 2018; Nopoulos, 2022) but alongside these developments there has been an unfavourable aspect to the history of the disease (Harper, 2002). In a historical narrative of the early American settlers in New York, the same area from which George Huntington drew his observations, Wexler (2002) found evidence of HD sufferers either being ostracised in communities or regarded as victims of witchcraft, and mitigation of the suffering caused by the disease in affected communities could only be achieved by social integration. A subsequent study by Wexler (2010) noted that at the turn of nineteenth century, the disease became more secret and hidden and this is due to wider societal changes rather than factors intrinsic to the disease.

EUGENICS AND THE ABUSE OF GENETICS

Since the first recognition of its Mendelian nature, HD has gained considerable attention (Harper, 1992; Wexler, 2012; Wexler, Wild and Tabrizi, 2016). Wexler (2012) observed that beyond the medical and scientific interest, it was also a source of interest of the social movement for better breeding known as Eugenics (Wexler, 2012) and became a cause for public health alarm (Wexler, 2002; Bombard et al., 2008). It was Charles Davenport, in collaboration with the physician Elisabeth Muncey (Davenport and Muncey, 1916) who published the first large scale study of HD, tracing 12 generations of affected families with a total of 4,370 individuals. Davenport and Muncey (1916) suggested that the prevention is the most obvious management of this 'dire inheritable disease' (p.215), and their publication was subsequently widely cited (Wexler, Wild and Tabrizi, 2016), providing the foundation for eugenicist arguments in favour of the proposition of voluntary sterilisation and immigration restrictions for 'persons with choreic lines' (Harper, 1992; Nance, 2017).

In his paper *HD and the abuse of genetics* (1992), Peter Harper outlined the long-established history of HD as the subject of considerable abuse in the nineteenth century. HD was notably at the centre of eugenic policies in Nazi Germany and other countries (Harper, 1992). Harper further stated that it would be unwise to assume that these attitudes to HD could not occur again in our current social system, a sentiment shared by Wexler (2012), given that, while it is unusual to blame stigma and discrimination on ignorance and misunderstanding, history shows that those with accurate knowledge of HD have also been perpetrators.

The historical reviews of Harper (1992, 2002) and Wexler (2012, 2016) suggested that by addressing the preceding impact of eugenics on HD, on individuals and families, HD research can help explain the misunderstanding, secrecy and shame that currently still surrounds the illness. The history of HD reminds us of the importance of recognising the experiences of the past and through developments in HD research has suggested ways to help us improve our understanding and care of those suffering from this devastating disease in the future.

THE HD GENE

HD is a neurodegenerative disorder governed by genetics (Nance, 2017; Pagan, Torres-Yaghi and Altshuler, 2017). HD is caused by a mutation in the gene by an abnormal protein referred to as a Huntingtin protein (HTT). As the mutant HTT protein accumulates, this negatively affects multiple cellular features (Pagan, Torres-Yaghi and Altshuler, 2017). A summary of the clinical understanding of HD is presented in Figure 2. The mutant gene produces an abnormal protein (HTT) which damages particular cells in the brain resulting in the clinical and pathological features of HD (Quarrell, 2008).

Figure 2. Summary of our understanding of HD (Quarrell, 2008)

HD can occur at all life stages, although disease onset before the age of 20 (Juvenile HD) or after 65 is relatively rare (Paulson and Albin, 2011). The mean age of HD onset is between 30 and 50 years (Roos, 2010). HD is transmitted in an autosomal dominant fashion (Figure 3), which means that males and females are equally affected, and that the offspring of an affected parent has a 50 percent chance of inheriting the disease mutation (Groves, 2017) . In 1983, Gusella and colleagues located the genetic marker in the short tip of chromosome 4 (Nance, 2017; Quaid, 2017). A decade after, the mutation causative of HD (HTT gene) was discovered by a group of HD investigators (Kay, Hayden and Leavitt, 2017). According to Testa and Jancovic (2019) these discoveries opened the 'gene hunter era' (p. 52). The continuing developments in the genetic aspect of HD has permitted the advent of gene-based research and therapies, to which researchers have referred as the dawn of a new era in HD (Nance, 2017; Testa and Jankovic, 2019).

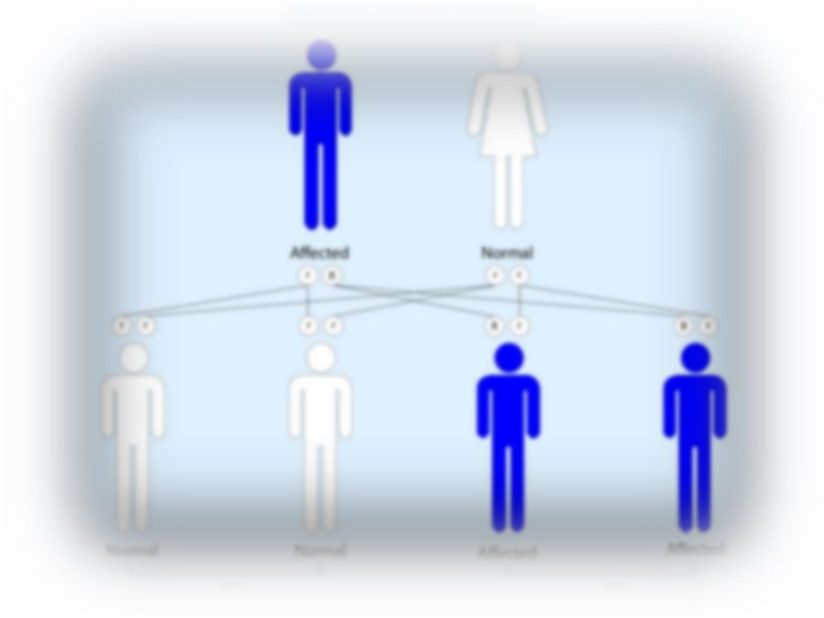


Figure 3. HD Autosomal Dominant Inheritance

Source: https://www.medicalexamprep.co.uk/mendelian-inheritance/autosomal-dominant/

CLINICAL DIAGNOSIS AND DISEASE STAGES

HD is diagnosed on the basis of clinical examination, familial history (if available) and DNA testing (Bates et al., 2015). The typical course of HD progression is divided into clinical pre-manifest and manifest stages. The pre-manifest stage is further subdivided into the pre-symptomatic phase, where individuals are clinically unaffected, and the prodromal phase, where individuals may present with subtle motor, cognitive or psychiatric symptoms (Pagan, Torres-Yaghi and Altshuler, 2017; Ghosh and Tabrizi, 2018). The manifest period is conventionally defined by the presence of a motor clinical symptom, usually the classic involuntary movement known as 'chorea'(Paulson and Albin, 2011). Chorea, and other movement disorders in HD, are further discussed in the succeeding subsection under the heading 'HD clinical features.'

Clinical diagnosis of HD manifest symptoms is based on the Unified Huntington's Disease Rating Scale (UHDRS). The UHDRS is the most widely used HD objective measure in clinical practice where clinicians assess the motor, cognitive, behavioural, and functional capacities present in HD. The clinical assessment of motor symptoms in HD is assessed through the UHDRS Total Motor Score (UHDRS TMS), this scale examines all features of HD movements, and the characteristic findings are usually strongly supportive of HD diagnosis(Bates et al., 2015).

HD diagnosis is a difficult and emotional period for affected individuals and families, as some have witnessed other members of their family who suffered the same fate and the affected persons are confronted with the inevitable debility (Ghosh and Tabrizi, 2018). In addition, the implications of the diagnosis can also affect their employment, driving and, for some countries, their insurance. Work and financial capacity are often the first areas of decline in people with early HD (Pagan, Torres-Yaghi and Altshuler, 2017). The disease progression from the onset is commonly illustrated through the five stages of HD. The clinically accepted stages of HD were originally described in 1979 by Shoulson and Fahn (Table 3), although a revised staging framework has been recently proposed in clinical research settings (Tabrizi et al., 2021). However, Shoulson and Fahn's staging system still remains widely used to date (Nopoulos, 2022).

Table 3. **HD stages, adopted from Shoulsun and Fahn (1979)**

HD stage	Occupation	Finances	Domestic Chores	Activities of Daily Living	Care Setting
Stage I (0-8 years)	Normal	Normal	Full	Full	Home
Stage II (3-13 y)	Reduced	Slight assistance	Full	Full	Home
Stage III (5-16 y)	Marginal	Major assistance	Impaired	Minimally impaired	Home
Stage IV (9-21 y)	Unable	Unable	Unable	Moderately impaired	Home or extended care facility
Stage V (11-26 y)	Unable	Unable	Unable	Severely impaired	Total care facility

The HD stages in clinical terms correspond to the Total Functional Capacity (TFC), which is a subscale in the UHDRS. This scale assesses the general function of HD individuals within five stages of functional domains (Shoulson and Fahn, 1979; Ghosh and Tabrizi, 2018). The TFC rating scale can be seen in Table 4. The five domains outline the general functional decline in HD, and with higher scores in each domain signifying better function. In clinical settings, the common term to describe the HD progression is early, middle, and late (Nopoulos, 2022).

Table 4. The TFC Score and its relationship to Shoulson and Fahn's stage and clinical descriptors, adapted from Ghosh and Tabrizi (2018)

TFC Domain	Level of Functioning	Score
Occupation	Normal	3
	Reduced capacity for usual job	2
	Marginal work only	1
	Unable	0
Finances	Normal	3
	Slight Assistance	2
	Major Assistance	1
	Unable	0
Domestic Chores	Normal	2
	Impaired	1
	Unable	0
Activities of Daily Living	Normal	3
	Minimal Impairment	2
	Gross tasks only	1
	Total care	0
Care Setting	Home	2

	Home with chronic care	1
	Full time skilled nursing care	0
HD Stage	**TFC Score**	
Early, Stage I	11-13	
Early, Stage II	7-10	
Middle, Stage III	4-6	
Late, Stage IV	1-3	
Late, Stage V	0	

HD Stage I (early) refers to TFC 11-13, stage II (early) refers to TFC 7-10, (middle) stage III refers to TFC 3-6, Stage IV (late) refers to TFC 1-2 and stage V (late/end) refers to TFC 0. Despite the usefulness of disease staging in clinical settings, researchers emphasise that the decline in functional capacity in HD individuals varies and that the disease clinical manifestations widely differ over the course of the illness progression (Pagan, Torres-Yaghi and Altshuler, 2017).

GENETIC TESTING

HD clinical manifestations are caused by mutations in the HD gene from the CAG repeats which lead to an expanded sequence in mutant HTT protein (Nance, 2017). The presence of extended CAG repeats became the widely accepted confirmation of HD for people with suggestive signs and symptoms of the disease (Kay, Hayden and Leavitt, 2017). Although neuroimaging and other tests can support the diagnosis, these are primarily used only to rule out other conditions and are usually unnecessary when a family history is confirmed followed by a positive genetic test (Bates et al., 2015). The relationship between genetic test results based on CAG repeats and patterns of hereditary risk and status is summarised in Table 5. HD studies suggest that the timing of disease onset and its severity are indicated by the number of expanded CAG (Kay, Hayden and Leavitt, 2017; Pagan, Torres-Yaghi and Altshuler, 2017). Higher numbers of repeats suggest the possibility of an earlier age of onset and severe declines in functioning. HD is fully penetrant if the CAG repeat is greater than or equal to 40, as this will always lead to HD and have a “positive” test result (Novak and Tabrizi, 2011; Nance, 2017). A “normal” HTT gene has less than 36 CAG repeats. Individuals with repeat lengths in the 27-35 range are termed “intermediate or indeterminate” and show subtle symptoms or may be unaffected (Pagan, Torres-Yaghi and Altshuler, 2017). Where the CAG repeat lengths range between 36 and 39 the result is termed “abnormal” and to reflect the reduced penetrance, and although this range is associated with HD, not everyone with these lengths will develop HD, and for

those who do develop the disease, the symptoms are most likely to manifest in later onset (Novak and Tabrizi, 2011).

Table 5. Clinical interpretation of CAG repeat lengths (Novak and Tabrizi, 2011; Nance, 2017)

CAG repeat length	Interpretation	HD status	Hereditary risk
10-26	Normal	Unaffected	None
27-35	Normal, Mutable	Unaffected	Increased but <50%
36-39	Abnormal, Reduced penetrance	May or may not be affected	50%
40 and above	Abnormal, Complete penetrance	Affected	50%

DNA testing is the current gold standard for determining an accurate HD diagnosis(Pagan, Torres-Yaghi and Altshuler, 2017). DNA or genetic testing is performed to measure the length of CAG repeats in the HTT gene, and testing falls into two categories: Diagnostic and Predictive. Diagnostic testing is undertaken to confirm or refute the diagnosis of symptoms suggestive of HD, while predictive testing is carried out with individuals without symptoms but who are at risk of HD due to their family history (Novak and Tabrizi, 2011).

EXPERIENCES OF GENETIC TESTING

Despite the availability of genetic testing, the uptake rates for individuals who are at risk of HD remains low (Rew et al., 2010; Quaid, 2017; Anderson et al., 2019). A survey carried out between 1999 and 2008 of nearly 800 individuals involved in the Prospective Huntington At-Risk Observational Study (PHAROS) found that the primary reasons for refusing to undergo genetic testing include the inability to undo knowledge and the lack of an HD cure or effective treatments (Anderson et al., 2019). Numerous research studies have explored the psychological consequences of genetic testing (Almqvist et al., 1999; Broadstock, Michie and Marteau, 2000; Decruyenaere et al., 2003; Rew et al., 2010; Hagberg, Bui and Winnberg, 2011; Smith et al., 2013; Andersson et al., 2016; Groves, 2017; Quaid, 2017). Although not limited to HD, Paulsen et al (2013) reviewed studies pertaining to HRQOL after predictive testing for neurodegenerative diseases and they suggested that although people reported no regrets and found extensive benefits in receiving genetic information, experiences of stigmatization and discrimination remain poorly understood. Williams et al (2010) explored narrative data from an HD international survey (I-RESPOND) and found that reports of stigma and genetic discrimination are both very personal and difficult to generalise, and that both interpersonal factors and policy context contribute to the outcome of potentially discriminating events.

Hopelessness was identified in individuals at high risk, and it has a long term consequence between HD carriers and non-carriers (Tibben et al., 1997). Earlier studies have associated genetic testing with intense distress, risks of psychiatric hospitalisation, suicide, and suicidality (Kessler, 1987; Almqvist et al., 1999). However, these claims were refuted by systematic reviews that established that the negative psychological impact of predictive testing was not associated with test outcomes (Broadstock, Michie and Marteau, 2000; Crozier, Robertson and Dale, 2015) and that catastrophic outcomes were rare (Paulsen et al., 2013). Crozier et al (2015) revealed that living with the HD gene carrier is not in itself clinically distressing. Qualitative studies further described a range of responses to undergoing a genetic test, with some positive and some negative effects (Etchegary, 2009; Hagberg, Bui and Winnberg, 2011; Groves, 2017). Hagberg et al (2011) noted that while some participants reported more positive family relationships and a greater appreciation of life, others felt they had experienced negative psychological effects and regretted their decision. Schwartz (2010) explored the narratives of individuals who received a positive HD diagnosis and noted that while some found purpose and meaning through diagnosis, others viewed suicide as a reasonable alternative to debilitating disability.

Groves (2017) emphasised that it is crucial to ensure that individuals seeking genetic tests fully understand the implications of their test results and how to use the diagnosis. Researchers suggest that testing protocols should include a pre-assessment of the individual's emotional state so that post-counselling testing and follow-up can be addressed to those who are more distressed (Broadstock, Michie and Marteau, 2000). HD guidelines recommend that testing protocols include neurologic examination, pre-test counselling, results in person and follow-up with assessment from psychiatrist or psychologist (Quaid, 2017). While guidelines suggest that the psychiatrist or psychologist can act as a gatekeeper in the testing process, some patient advocacy groups have criticised this as "hurdles to pass through" and believe that receiving a test result is an individual's right (Groves, 2017). Groves (2017) recommends that clinicians should find a balance between the ethical principles of autonomy and should explore the individual's motivation for genetic testing to ensure that appropriate support is offered. These findings were supported by an earlier study that found that post-test psychological distress can be associated with poorly developed social support and lack of psychological defensive strategies (Kessler, 1987). Similarly, a longitudinal study by Decruyenaere et al., (2003) suggests that counselling should pay attention to persons with lower ego strength and with unspecified test motivation, as these characteristics carry a high risk of distress independent of test results. While MDT involvement, in particular counselling in HD pre- and post- genetic results, is warranted, more research is needed to establish the type of support, and the extent of psychological provisions, that HD services should offer (Crozier, Robertson and Dale, 2015).

EPIDEMIOLOGY

The hallmark of modern epidemiological measures in HD is the confirmation of CAG genetic repeat (Bates et al., 2015). Prevalence estimates depend on comprehensive genetic testing and on examination of clinical signs to accurately assess disease onset (Bates et al., 2015; Kay, Hayden and Leavitt, 2017). Reviews of HD

epidemiology suggest a wide variation of disease prevalence (Pringsheim et al., 2012; Rawlins et al., 2016) within populations of different ancestry (Baig, Strong and Quarrell, 2016; Kay, Hayden and Leavitt, 2017).

Although HD is endemic to all populations, the highest prevalence is seen among Caucasians, and the lowest in Asians (Rawlins et al., 2016). The average prevalence rate is 5.70 per 100,000 in Europe, Australia and North America (Pringsheim et al., 2012). In Japan prevalence is recorded at 0.72 per 100,000 with much lower incidence in Taiwan and Hong Kong (Kay, Hayden and Leavitt, 2017). African populations show a similar reduced prevalence, but there is higher frequency among individuals with European ancestry (Baig, Strong and Quarrell, 2016). Based on a systematic review , the worldwide prevalence of HD is 2.71 per 100,000 (Pringsheim et al., 2012).

HD prevalence is widely studied in the UK (Kay, Hayden and Leavitt, 2017), with data enumerated from GPs, hospitals and nursing home records (Evans et al., 2013). A study using reports from General Practice Research Databases (GPRD) showed an increase in UK prevalence from 5.4 in 1990 to 12.3 in 2010. Wexler et al. (2016) reported that despite the rise of prevalence in the UK, the incidence rate remained constant (Wexler et al., 2016). It is unclear if the increase in prevalence reported reflects the improved accuracy of surveys or an actual increase in HD prevalence in the intervening period (Pringsheim et al., 2012). Besides the UK, epidemiological studies have focused interest on the isolated cases in Tasmania and around Lake Maracaibo in Venezuela (Kay, Hayden and Leavitt, 2017). The concentrated focus of HD cases in these regions was believed to originate in a few founders of European lineage (Wexler et al. 2004). Research has identified that the difference in prevalence between ethnic groups relates to genetic differences in the HTT gene, it has also been observed that populations with high prevalence have longer average CAG repeats (Baig, Strong and Quarrell, 2016; Kay, Hayden and Leavitt, 2017). HD prevalence has increased up to 20 percent per decade (Rawlins et al., 2016; Wexler et al., 2016), and this could be attributed to longer lifespans and ageing populations (Evans et al., 2013; Kay, Hayden and Leavitt, 2017).

Despite the increase in worldwide prevalence, HD is considered as a rare disease. The European Union defined a rare disease as one that is progressive or life-threatening with a prevalence of less than 50 cases per 100, 000 (Spinney, 2010). However, acknowledgement of the genetic nature of HD clearly implies that the multiple generations of families have suffered from its consequences. Often, a number of individuals within the family are afflicted simultaneously, further compounding caregiving responsibilities (Parekh, Praetorius and Nordberg, 2017). If HD families and caregivers are counted, this devastating disease affects a considerable number of people (Quarrell, 2008).

THE CLINICAL FEATURES OF HD

By the late 1900s, the term Huntington's Chorea had been replaced in medical literature by the more inclusive term, Huntington's disease (HD). This change is an acknowledgement that HD is a complex disease, and that chorea is just one of its features (McCusker and Loy, 2017). Although HD is mainly classed as a movement disorder, the associated behavioural and cognitive impairments also place it within

progressive neurological dementias and neuropsychiatric disorders (Testa and Jankovic, 2019). HD is characterised by a triadic syndrome of motor, psychiatric and cognitive impairments (Figure 4), referred to as a 'unique constellation of HD signs and symptoms' (Ho et al., 2009).

The symptoms of the disease intensify over 15 to 20 years' duration from diagnosis through complete loss of independence to death. Aspiration pneumonia and suicide are the most common causes of death (Wexler et al., 2016; Patrick and Ritchie, 2020). To date, available treatments for HD centre on management of the disease symptoms, and despite the concerted scientific efforts, and some promising experimental HD therapies, a cure in HD remains elusive (Wild, 2016; Testa and Jankovic, 2019).

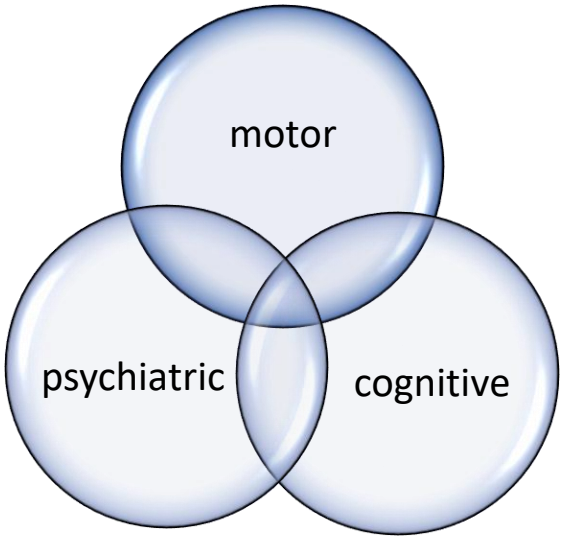


Figure 4. The HD triad: motor, cognitive and psychiatric symptoms

THE HD BRAIN

The symptoms of HD are linked to the neurodegeneration of the brain. The HTT protein causes damage to the neuronal cells in the brain, particularly in the cerebral cortex and the basal ganglia (van der Burg, Björkqvist and Brundin, 2009; Waldvogel et al., 2014). Walker (2007) noted that prominent cell loss and atrophy have been found in selective areas in the caudate nucleus and putamen (collectively known as striatum). The striatum is the major input to the basal ganglia which are involved in the brain circuits that, aside from controlling motor movements, also mediate cognitive and affective functions (Waldvogel et al., 2014). This striatal loss, which involves the medium spiny neurons of the brain is considered the pathological hallmark of HD (Testa and Jankovic, 2019). A study by Waldvogel et. al. (2014) found that the HD brain exhibited extensive striatal loss in basal ganglia when compared with healthy controls (Figure 5). These neuropathological changes in the HD brain have been linked to its classic motor, psychiatric and cognitive disease features (van der Burg, Björkqvist and Brundin, 2009).

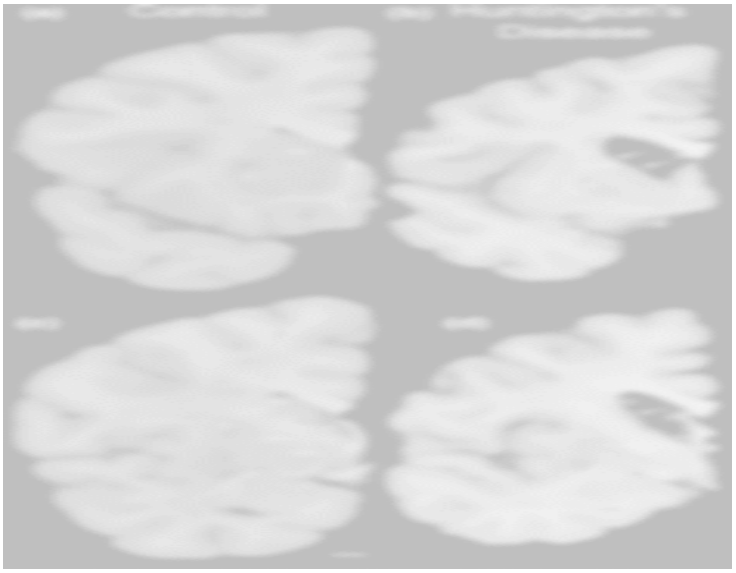


Figure 5. Pathology of HD diseased brain (Waldvogel et al., 2014)

Legend: Coronal sections of brain of a) and c) in control case and b) and d) in HD case showing the difference in striatum of the basal ganglia regions. The arrows in the scans indicated a major shrinkage caused by cell loss in the caudate nucleus and putamen (Waldvogel et al., 2014)

MOTOR FEATURES: CHOREA AND OTHER MOVEMENT DISORDERS

The primary degeneration in the basal ganglia classified HD as a disease of movement disorders. There is a wide range of motor manifestations present in HD. This review has summarised the motor features identified in the literature search, and these can be seen in Table 6. While some motor disorders are more prominent than others, chorea is the most distinguishable feature of HD (McGarry and Biglan, 2017; Sherman et al., 2019). Chorea, a term derived from an ancient Greek word that means dance, often begins as a fleeting suppressible movement that may appear as fidgetiness (Paulson and Albin, 2011). In a focus group study, Sherman et al (2019) found that living with chorea significantly impacts the quality of life of affected individuals across the HD spectrum. Studies have documented that people with HD are often unaware of the presence of chorea (Sitek et al., 2011; Sherman et al., 2019), and individuals are more concerned by the interference in voluntary activities such as writing, eating and walking (Ghosh and Tabrizi, 2018). Moreover, people with HD are often accused of either nervousness or drunkenness by people unaware of their diagnosis (Roos, 2010; Ghosh and Tabrizi, 2018). The common reasons for seeking medical interventions for chorea include embarrassment, stigma and physical injuries resulting from the symptom (Burgunder et al., 2011).

Table 6. List of Motor Symptoms identified from the literature review

HD motor disorders	Definition
Akathisia (Bachoud-Lévi et al., 2019)	Inability to stay still
Akinesia (Roos, 2010; Frank, 2014; McCusker and Loy, 2017; Ghosh and Tabrizi, 2018)	Difficulty in initiating movements
Apraxia (McCusker and Loy, 2017; Petersén and Weydt, 2019)	Difficulty with motor planning and performing tasks
Ataxia (Roos, 2010)	Lack of voluntary coordination of muscle movements that can include gait abnormality, speech changes, and abnormalities in eye movements
Athethosis (Petersén and Weydt, 2019)	Slow, writhing, continuous, involuntary movements
Ballism (Frank, 2014; Bachoud-Lévi et al., 2019)	Repetitive, but constantly varying, large amplitude involuntary movements of the proximal parts of the limbs
Bradykinesia (Novak and Tabrizi, 2011; Frank, 2014; Ghosh and Tabrizi, 2018; Petersén and Weydt, 2019; Testa and Jankovic, 2019)	Slowness of movement
Bruxism (Novak and Tabrizi, 2011; Bachoud-Lévi et al., 2019)	Grinding or clenching of teeth
Chorea (McCusker and Loy, 2017; Testa and Jankovic, 2019; McGarry and Biglan, 2017; Novak and Tabrizi, 2011; Roos, 2010, Bachoud-Lévi et al., 2019; Mestre and Shannon, 2017; Ghosh and Tabrizi, 2018; Frank, 2014; Sherman et al., 2019; Petersén and Weydt, 2019)	Involuntary, irregular, purposeless, non-rhythmic, abrupt or rapid movements that seem to flow from one body part to another
Clonus (McCusker and Loy, 2017)	Muscular spasm involving repeated, often rhythmic, contractions
Dysarthria (Roos, 2010; McCusker and Loy, 2017; Bachoud-Lévi et al., 2019; Petersén and Weydt, 2019; Testa and Jankovic, 2019)	Difficulty in speech due to weakness of speech muscles
Dysphagia (Roos, 2010; Frank, 2014; McCusker and Loy, 2017; Ghosh and Tabrizi, 2018;	Difficulty in swallowing

Bachoud-Lévi et al., 2019; Petersén and Weydt, 2019)	
Dystonia (Novak and Tabrizi, 2011; McGarry and Biglan, 2017; Testa and Jankovic, 2019; McCusker and Loy, 2017; Roos, 2010; Bachoud-Lévi et al., 2019; Ghosh and Tabrizi, 2018; Frank, 2014; Petersén and Weydt, 2019)	Twisting movements and postures that tend to be sustained at the peak of the movement, often patterned and repetitive
Hyperkinesia (Roos, 2010; McCusker and Loy, 2017; Petersén and Weydt, 2019)	An increase in bodily movement
Hyperreflexia (Roos, 2010)	Overactive or over responsive reflexes
Hypokinesia (Roos, 2010; McCusker and Loy, 2017; Petersén and Weydt, 2019)	A decrease in bodily movement
Myclonic jerks (Novak and Tabrizi, 2011; Frank, 2014; McCusker and Loy, 2017; Ghosh and Tabrizi, 2018)	Sudden, brief, shock-like involuntary movements caused by muscle contractions or sudden loss of muscle tone
Opisthonus (Ghosh and Tabrizi, 2018)	Spasm of the muscles causing backward arching of the head, neck, and spine
Rigidity (Roos, 2010; Novak and Tabrizi, 2011; Ghosh and Tabrizi, 2018; Bachoud-Lévi et al., 2019; Testa and Jankovic, 2019)	Increased muscle tone which is felt when the examiner tries to move the neck, arm or leg when completely relaxed
Spasticity (Novak and Tabrizi, 2011)	Tight or stiff muscles and an inability to control those muscles
Tics (Roos, 2010; Frank, 2014; McCusker and Loy, 2017; Ghosh and Tabrizi, 2018; Petersén and Weydt, 2019)	Fast, repetitive muscle movements that result in sudden and difficult-to-control body jolts or sounds
Torticollis (Roos, 2010; Ghosh and Tabrizi, 2018)	Focal dystonia of the neck muscles in which there is a simultaneous contraction of opposing muscle groups causing the neck to twist, tilt, shift, and pull forwards or backwards
Tremors (McCusker and Loy, 2017)	Oscillation that is usually rhythmical and regular and that affects one or more body parts

Motor impairments in HD can be broadly classified in two phases: the hyperkinetic and hypokinetic (McColgan and Tabrizi, 2018). Over time as the disease progresses, hyperkinetic movements with the prominence of chorea are observed to lessen and replaced by other hypokinetic symptoms such as dystonia (a common feature in younger onset), bradykinesia, rigidity and impaired postural reflexes (Novak

and Tabrizi, 2011; Bachoud-Lévi et al., 2019; Testa and Jankovic, 2019). People with HD can experience involuntary movements that cause coordination problems, gait disturbances, and increases the risk of falls (Mestre and Shannon, 2017, Ghosh and Tabrizi, 2018). Bruxism, ballism and tremors are rare, yet distressing symptoms and these are not commonly recorded in clinical settings (McCusker and Loy, 2017). Other motor features include tics, myoclonus and abnormal postures such as torticollis and opisthonus (Ghosh and Tabrizi, 2018). The HD motor features are often related with other more complex neurological symptoms such as difficulty in swallowing (dysphagia), speaking (dysarthria), impaired eye movements and other executive motor functions (Petersén and Weydt, 2019). All of these affect the individuals' daily function, resulting in increased physical disability and reduced quality of life (Petersén and Weydt, 2019; Testa and Jankovic, 2019).

NON-MOTOR FEATURES: PSYCHIATRIC AND COGNITIVE SYMPTOMS

Although HD is predominantly classed as a movement disorder (Marsden, 1986), it also firmly connects the field of psychiatry and neurology. Wexler et al. (2016) observed that while some neurologists claim HD as a 'neurological disease par excellence', many more psychiatrists deal with HD in psychiatric institutions. Paulson and Albin (2011) therefore argued that HD in every case can be seen as a 'neuropsychiatric disorder'. While the motor symptoms of HD are the most immediately apparent, non-motor symptoms generate the greatest burden on individuals and families (Craufurd and Snowden, 2002; Paulson and Albin, 2011; Simpson et al., 2016) and constitute a key precursor to the loss of independence (Craufurd and Snowden, 2002). Moreover, it is these symptoms that cause the most distress and are often the cause of hospitalization (Patrick and Ritchie, 2020).

PSYCHIATRIC SYMPTOMS

As in the case of motor features, the range of neuropsychiatric symptoms occurring in HD is broad. Studies identified that depression, aggression, obsessive compulsive behaviour, psychosis, and apathy are highly prevalent in the HD population (Craufurd, Thompson and Snowden, 2001; van Duijn, 2017). The prevalence of these symptoms in relation to the HD stages can be seen in Figure 6. Psychosis, while serious, is a relatively rare symptom with prevalence as low as 1% (McColgan and Tabrizi 2018), and this mainly appears in the later stages of the disease.

Figure 6. Prevalence of moderate to severe symptoms on the behavioural subscales across different disease stages. (van Duijn et al., 2014)

Legend: Disease stages were defined according to the Total Functional Capacity (TFC) score: Stage 1=TFC score 13-11, Stage 2=TFC score 10-7, Stage 3=TFC score 6-3, Stage 4=TFC score 2-1, Stage 5 =TFC score 0. Due to the low number of mutation carriers in Stage 5 (n=23), Stages 4 and 5 were combined. (van Duijn et al., 2014)

Of all psychiatric features, depression is the most prominent, with studies documenting the prevalence as high as 76% (Patrick and Ritchie 2020). Depression in HD can be difficult to diagnose (Crauford and Snowden 2002) as people with HD may not seek interventions for sadness or low mood, and such symptoms can be masked by other disease features such as weight loss, inactivity, or social withdrawal (Roos, 2010). Depression in the HD population is not fully understood (Paulsen et al., 2017; McAllister et al., 2021), as this can be a presenting symptom or in overt disease (Mestre and Ferreira, 2012). Studies have documented that depression is more common in the early stages of the disease (Paulsen et al., 2017) and this declines as the disease progresses (van Duijn et al., 2014). High levels of depression have been found when individuals undergo genetic testing, and prior to receiving a formal diagnosis, but the possibility cannot be ruled out that these symptoms relate to a perception of stress during what is necessarily a period of uncertainty (Paulsen et al., 2017).

Depression is seen amongst those with HD at twice the rate of the general population and over 10 percent have made a suicide attempt (Paulsen et al 2005, Patrick and Ritchie 2020). The rate of suicide is higher in cases of HD than in other neurological conditions (Halpin, 2012) and is a major concern for individuals,

families and clinicians (Paulsen et al., 2017). Although suicidal ideation is present throughout the disease progression (Kachian et al., 2019) , the most crucial periods occur when the person with HD receives a positive diagnosis and when their level of independence decreases (Testa and Jankovic 2019). Halpin (2012) argued that while some studies asserted that suicides in HD are the result of pathophysiological changes, an individual can make a conscious decision to terminate life as a response to the realities of living with HD. Studies also found that the higher risk of suicidality is associated with depression, anxiety, previous suicide attempts and aggression (Kachian et al. 2019).

A systematic review of the rates of aggression in those with HD revealed that its prevalence is up to 66 per cent and is more common in males (Fisher et al 2014). In a study conducted with ten HD patients in inpatient rehabilitation settings, aggression incidents were high, and commonly occurred during personal care activities (Brown, Sewell and Fisher, 2017). Aggression (verbal and/or physical) directed towards families or friends can have damaging consequences on personal relationships, can contribute to marital breakdown, or can result in refusal of care by family members (Craufurd, Thompson and Snowden, 2001). Aggression and irritability may occur early in the disease process, while obsessions and compulsions are characteristics of the HD motor manifest stages (Testa and Jankovic, 2019). Although aggression is a common behavioural sequela in HD, further research is needed to identify the antecedent and precursor "triggers" of this disease manifestation (Fisher et al., 2014). Apathy, a key neuropsychiatric symptom occurs in about 70 per cent of the HD population (Krishnamoorthy and Craufurd, 2011). Apathy is a challenging symptom, characterized by general loss of interest and reduced goal-directed behaviour. In the European registry cohort study, apathy had the strongest inverse correlation with the disease stage progression, and the most frequent symptom documented in behavioural scales (van Duijn et al., 2014). Apathy contributes to HD personality changes (Petersén and Weydt, 2019), and is correlated with HD cognitive impairment (Andrews et al 2020). Whilst apathy, aggression and depression are all related to functional decline, apathy has been observed to be a symptom of the debilitating effect of the duration of the disease(van Duijn et al., 2014). Similarly, in a study where 134 HD patients were assessed using the Problem Behaviours Assessment for Huntington Disease (PBA-HD), an instrument for rating the presence, severity and frequency of behavioural abnormalities in HD, it was found that apathy was highly correlated with illness duration (Craufurd, Thompson and Snowden, 2001). McGolgan and Tabrizi (2018) argued this can be attributed to the lack of effective treatments for apathy in comparison with the range of anti-psychotics and anti-depressants used to address symptoms of depression, aggression, and anxiety.

COGNITIVE SYMPTOMS

Cognitive dysfunction is an invariable feature of HD, and has been observed early in the course of the disease (Craufurd and Snowden, 2002; Patrick and Ritchie, 2020). HD cognitive symptoms are progressive, which impacts executive functions, attention, and psychomotor speed, and over time leads to dementia (Testa and Jankovic, 2019). HD has been associated with changes in multiple brain regions, thus the cognitive manifestations encompass a wide variety of skills. Early cognitive changes include difficulty in multi-tasking, reduced concentration and diminished visuospatial functions, as well as impaired emotional

recognition (Novak and Tabrizi, 2011; Petersén and Weydt, 2019). Language and memory are impacted but in contrast to Alzheimer's dementia, these areas are relatively preserved and decline gradually in comparison to other cognitive domains (Testa and Jankovic, 2019). Anosognosia, described as lack of awareness of disease symptoms is commonly observed in HD (Sitek et al., 2011). Self-awareness is impaired in at least one third of people with HD (Paulsen et al., 2017), and this significantly affects clinical interventions as HCPs are unable to assess the breadth and impact of symptoms, especially when families' or caregivers' input is unavailable (Testa and Jankovic, 2019). Bourne et al (2004) reported that HD cognitive dysfunction often leads to behavioural problems and that adopting strategies to deal with the cognitive and behavioural deficits can be necessary (Table 7). Researchers argue that the distinction with HD neuropsychiatric symptoms is not always straightforward, as many individuals may also present disinhibition, difficulties in initiation, lack of awareness, and poor disease insight (Duff et al., 2007).

Table 7. Behavioural difficulties associated with HD cognitive problems(Bourne et al., 2006)

Behaviour	Possible management strategies
Fixed thinking	Try to channel fixed behaviour thought into a manageable and appropriate behaviour.
Slowed thinking and Responses	Use clear and concise sentences, use closed ended questions to elicit a specific response.
Memory problems	Use prompts, cues and memory aids. Apply step by step approach to the task and reduce environmental distractions.
Difficulty sequencing Activities	Maintain regular routines, use prompts and supervisions when required
Loss of visuospatial Awareness	This can be misinterpreted as aggressive behaviour, due to lack of insight into individual personal space. Take a step back from the person, environmental safety modifications i.e. flat flooring for safe mobilisation.

The cognitive aspect of HD is well studied in quantitative studies (Beglinger et al., 2010; Paulsen et al., 2017), and some of these large-scale studies will be discussed in the following section. The large multi-site longitudinal and observational studies (TRACK-HD and PREDICT HD) which identify biomarkers and quantify possible outcomes in HD progression, found that cognitive deterioration is present even in individuals who are at risk but not officially diagnosed with motor symptoms (Beglinger et al., 2010; Paulsen et al., 2017). Although studies have well documented the cognitive problems, including domains of complex executive functions in HD (Beglinger et al., 2010), a systematic review by Mestre et al (2018) identified that current HD cognitive scales needs to be validated. Similarly, Paulsen et al (2017) argued that future research needs

to emphasize the validation of current cognitive, behavioural, and functional assessments in clinical trials of individuals with early HD.

OTHER SYMPTOMS: SLEEP, METABOLIC CHANGES AND SEXUAL DISORDERS

In addition to the established motor, cognitive and psychiatric signs and symptoms, sleep disturbances are common in HD (Bachoud-Lévi et al., 2019) and have been linked with depression and cognitive decline (Aziz et al., 2010). Anxiety and night-time chorea are contributing factors to insomnia (Ghosh and Tabrizi, 2018). Bachoud-Levi et al (2019) noted that problems in diurnal rhythm (day-night reversal, etc.) are more common compared to simple insomnia. Attention has also been paid to the unintended weight loss associated with HD (Roos, 2010). Progressive weight loss is universal in the HD population, and this was previously linked to motor disorders and swallowing dysfunction (Kirkwood et al., 2001), however, other genetic studies found that there is a link with HD gene expansion (Aziz et al., 2008). Some researchers postulate that weight loss is multi-factorial, that the increased metabolic rate inherent to HD and other practical factors such as slower functioning, reduced appetite, and difficulty in food handling and swallowing can all be causative factors (Roos, 2010; Testa and Jankovic, 2019). Some would argue that peripheral issues of HD such as weight loss could be directly caused by the abnormal HTT protein and might occur independently from the neurological disease pathology (Waldvogel et al., 2014). Regardless of differing views seen in studies of sleep and circadian functions in HD, disturbances to sleep patterns remain an integral part of the disease feature, and have a substantial impact upon the HD individual's quality of life (Petersén and Weydt, 2019).

HD has also been associated with sexual disorders, although few studies have explored sexuality and sexual dysfunction in HD (Reininghaus and Lackner, 2015). A review by Reininghaus and Lackner (2015) found that sexual disorders such as increased sexual interests and paraphilia were common in HD. Crauford et al (2011) associated the hypersexual behaviours with other behavioural changes such as perseveration and obsessive-compulsive disorders. The mechanisms that affect these sexual abnormalities in HD are unclear. Reininghaus and Lackner (2015) argued that there is no evidence that sexual dysfunction is a specific symptom of HD but instead can be associated with the psychosocial factors related to brain changes brought about by pathological deterioration. Research on the extent of this possible association of sexual dysfunction with depression, irritability and dementia symptoms is needed to better understand the reasons for the sexual problems experienced in HD (Reininghaus and Lackner, 2015).

NATURAL HISTORY STUDIES AND FUTURE RESEARCH

A number of multi-centre natural history studies illuminated our understanding of the motor, psychiatric and cognitive variables in HD, particularly on the clinical aspect of the disease and imaging biomarkers (McColgan and Tabrizi, 2018). Table 8 presents details of some of these large-scale natural history studies in HD and their corresponding intervention goals. In addition, other promising HD clinical studies include GENERATION HD1, which is the first human research to test the HTT lowering therapy and has now entered

the phase three trial in 2019, and the PACE HD, which summarises evidence in physiotherapy and exercise in HD (Rodrigues, Quinn and Wild, 2019). These developments in HD research have helped us understand the disease pathogenesis and develop approaches to clinical treatment. Individuals affected with HD have the same underlying problem, and this is caused by the expanded CAG mutation in HTT (Figure 2.5). This monogenic nature and complete penetrance of HD should imply that disease-modifying therapies should be within reach, however such treatments do not currently exist (Mestre and Shannon, 2017; Pagan, Torres-Yaghi and Altshuler, 2017). HD remains a complex disease and is referred to as the 'most curable incurable brain disorder (Wild, 2016).'

Table 8. Examples of large scale and natural history studies in HD

STUDY NAME	STUDY DESIGN AND CHARACTERISTICS
COOPERATIVE HD OBSERVATION RESEARCH TRIAL (COHORT)	A prospective study organised by the international Huntington Study Group that collected data from yearly assessments and related clinical characteristics (phenotypes) to genetic and environmental factors. https://clinicaltrials.gov/ct2/show/NCT00313495 (Dorsey, 2012)
PROSPECTIVE HUNTINGTON AT RISK OBSERVATIONAL STUDY (PHAROS)	The study involved over 1,000 participants with 42 clinical sites based in the US. The prospective study monitors individuals who are at risk of developing HD and tracks changes using the UHRDS clinical assessment tool. https://clinicaltrials.gov/ct2/show/NCT00052143 (Anderson et al., 2019)
PREDICT HD	The study included over 1,400 worldwide participants at risk of HD. The focus of the study is to predict the earliest signs of HD. https://clinicaltrials.gov/ct2/show/NCT00051324 (Paulsen et al., 2014)
TRACK HD/ TRACK-ON HD	The study includes a total of 366 participants consisting of controls, pre-manifest and early HD. The study has 4 sites in different countries and focuses on evaluation and identification of early HD biomarkers for clinical trials. https://portal.dementiasplatform.uk/CohortDirectory/Item?fingerPrintID=TRACK%20HD (Tabrizi et al., 2012)

ENROLL HD **(FORMERLY CALLED REGISTRY)**	Worldwide observational studies for HD families. The research currently involves 19,734 participants, 178 clinical sites with 20 nations participating. The goal of the research is to accelerate new therapeutics in HD. https://www.enroll-hd.org/ (Landwehrmeyer et al., 2017)

TREATMENT AND CLINICAL CARE

Currently, there is no treatment that can slow the disease progression (Patrick and Ritchie, 2020; Nopoulos, 2022). HD care focuses on medical and non-medical management of symptoms provided by a Multidisciplinary care team (MDT). Variable disease symptoms can improve with pharmacological and behavioural interventions (Walker, 2007; Petersén and Weydt, 2019). Although HD is incurable, the disease itself is not untreatable (Wexler, Wild and Tabrizi, 2016).

PHARMACOLOGICAL INTERVENTIONS

To help clinicians manage the plethora of HD symptoms, numerous guidelines and clinical reviews have been published over the past decade (Roos, 2010; Novak and Tabrizi, 2011; Frank, 2014; Bates et al., 2015; McCusker and Loy, 2017; Mestre and Shannon, 2017; McColgan and Tabrizi, 2018; Petersén and Weydt, 2019; Testa and Jankovic, 2019; Patrick and Ritchie, 2020), including the collaborative work of international HD experts commissioned by the European Huntington's Disease Network (EHDN) (Bachoud-Lévi et al., 2019). These guidelines have included pharmacological and non-pharmacological management of HD symptoms. An example of HD treatment algorithm can be seen in Figure 7.

Chorea is the only symptom which can currently be treated (Mestre and Shannon, 2017). To date, there are only two approved US Food and Drug Administration drugs to treat HD symptoms, these are dopamine depleting agents namely tetrabenaxine and deutetrabenaxine, which are primarily used to treat chorea (Petersén and Weydt, 2019). Treatment of chorea is important to address both function and QOL of people with HD (Ghosh and Tabrizi, 2018). However, in a survey of 3,600 respondents, nearly 40% reported not utilising any medications to treat chorea, and nearly 25% reported that the available medications are unable to effectively suppress the symptom (Simpson et al 2016). McCusker and Loy (2017) argued that treatment needs to be in the context of the whole disorder, as medications may aggravate or adversely

impact other symptoms. For example, treating a person with chorea, dysphagia and depression, with dopamine depleting drugs may worsen their depression and further degrade swallowing (McCusker and Loy, 2017). Frank (2014) noted that the pharmacological rationale for treatment of chorea is not clear, and the observed symptomatic outcomes are related to non-specific effects, with sedation as the most common effect.

HD guidelines exist on the treatment of symptoms like depression, anxiety, and psychosis (van Duijn et al., 2014; Bachoud-Lévi et al., 2019), but for more disease-specific symptoms like apathy, irritability, and obsessive-compulsive behaviours, off-label use of psychotropic medications is unavoidable since evidence for all interventions in HD remains limited (van Duijn, 2017). The lack of evidence in treating depression in HD is of particular concern, given the increased risk of suicidality (Patrick and Ritchie, 2020). RCTs of pharmaceutical drugs found little evidence of their efficacy in treating HD cognitive symptoms (McColgan and Tabrizi, 2018). Patrick and Ritchie (2020) suggested a treatment algorithm for specific HD symptoms, distinguishing between physical and social issues (Figure). The goal of any treatment should be acceptability to patients, families and the treating clinicians (Frank, 2014). A Cochrane systematic review concluded that further drug trials with greater methodological quality should be conducted and future studies should include early stage, non-symptomatic HD patients (Mestre et al., 2009). Although, several HD features may be treated, the evidence for their effectiveness is lacking (Testa and Jankovic, 2019), and there is a need for better, disease modifying drugs (Roos, 2010). High-quality evidence for the treatment of HD in general remains limited. Although HD treatments exist, these are mostly based on expert consensus (Bachoud-Lévi et al., 2019; Patrick and Ritchie, 2020).

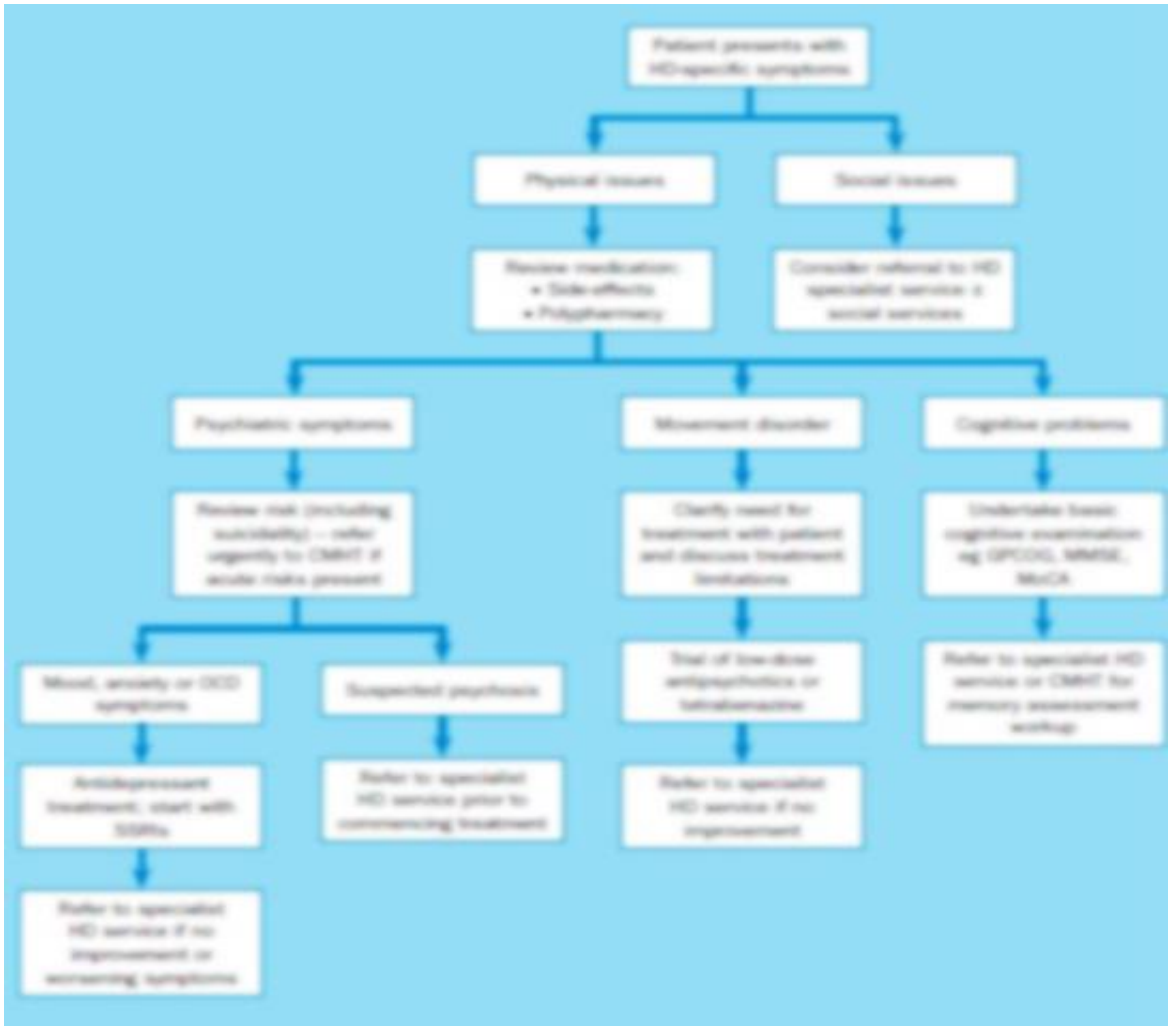


Figure 7. Treatment algorithm for HD patients presenting with specific symptoms (Patrick and Ritchie, 2020)

NON-PHARMACOLOGICAL INTERVENTIONS

In addition to drug therapies, non-pharmacological interventions may prove beneficial in addressing symptoms in HD and may help improve quality of life (Bachoud-Lévi et al., 2019). Novak and Tabrizi (2010) reported that non-drug-based measures are often more helpful in managing the disease features. Examples of such management therapies can be seen in Table 9. The non-pharmacologic interventions may come before the prescription of medication or can be provided in conjunction (van Duijn, 2017). Several clinical guidelines are also available to support allied health professionals in OT, PT, communication, nutrition, and SALT. HD has been shown to be modulated by environmental factors such as physical activity, cognitive stimulation, and diet (Mestre et al., 2009; Bachoud-Lévi et al., 2019). Preliminary studies have also suggested the potential therapeutic benefits of music, rhythm and dance on the symptoms of people with HD (Schwartz et al., 2019). Exercise therapies have been extensively researched and have reported a range of subjective physical and social benefits (Fritz et al., 2017; Mueller, Petersen and Jung, 2019; Rodrigues, Quinn and Wild, 2019). A systematic review by Mueller et al. (2019) found that exercise has a beneficial effect on cardiovascular and mitochondrial function in the HD population, although the effects on cognition, motor function, and body composition are inconsistent. Moreover, based on other

data from progressive neurological diseases, cognitive behavioural interventions may also be useful with HD (Fernie, Kollmann and Brown, 2015). Lifestyle factors such as education, occupation and engagement in brain-challenging activities can delay the onset of HD symptoms and improve cognitive performance (Garcia-Gorro et al., 2019). Dietary interventions such as nutritional supplements have been shown to meet the disease metabolic demands (Trejo et al., 2005) and Mediterranean food has been associated with better QOL in HD cohorts (Rivadeneyra et al., 2016). In the absence of effective treatments to slow the course of HD, lifestyle interventions offer feasible approaches to delay the disease, and can be combined with pharmacological therapies (Mo, Hannan and Renoir, 2015).

Table 9. Examples of non-pharmacological management in HD (Novak and Tabrizi, 2011)

Feature of disease	Management examples
Gait disturbance and chorea	PT to optimise and strengthen gait and balance. OT assessment to modify home environment and improve safety.
Cognitive symptoms	Ensure each day has a structure to overcome apathy and difficulty in initiating activities. Seek advice from OT. Maintain routines to reduce need for flexibility.
Social Problems	Caregivers to help at home. Residential settings or day centres to maintain social interactions.
Communication	SALT to optmise speech and assess for aids. Ensure information is presented simply, and allow time to comprehend and respond to speech.
Nutrition	SALT to advise on safe food consistencies at different HD stages. During the later stages, consider advice on enteral nutrition. Dietician to optimise nutritional intake. Minimise distractions to optimize swallowing safety.
Psychological Problems	Develop strategies to deal with the cognitive and emotional challenges of the disease using counselling or cognitive behavioural therapies.

Sufficient evidence is needed to support the neuroprotective effects of these non-drug therapies in people with HD. Systematic reviews concluded that a strong evidence base is lacking (Bilney, Morris and Perry, 2003; Patrick and Ritchie, 2020), and most research is conducted in small feasibility studies (Mestre and Shannon, 2017; Mueller, Petersen and Jung, 2019). Recommendations on lifestyle modifications in HD

populations remain non-specific as there are no clear known environmental risk modifiers for HD onset or progression (Testa and Jankovic 2019). Future research must put emphasis on therapy outcomes (Bilney, Morris and Perry, 2003), utilise larger HD cohorts and long term interventions (Mueller, Petersen and Jung, 2019).

MULTIDISCIPLINARY CARE

In the absence of disease-modifying therapies and the poor evidence base for treatments in HD, a coordinated health approach is vital in the care of people with HD (Edmondson and Goodman, 2017; Mestre and Shannon, 2017). As is the case in complex chronic diseases such as PD (Qamar et al., 2017) and progressive dementias (Grand, Caspar and Macdonald, 2011), multidisciplinary care is considered the gold standard of care delivery (Frich et al., 2016). In this model (Figure 8), a team of professionals from different disciplines work together to provide comprehensive care and support to people affected by HD (Edmondson and Goodman, 2017). Located in a specialized HD centre, the team would include medical specialists from neurology, psychiatry and palliative care, alongside genetic counsellors, specialized HD nurses and allied health professionals such as PT, OT, SALT, and social workers (Mestre and Shannon, 2017). Nance and Westphal (2002) acknowledged the unique contribution of HD centres, and that some services may require input from advocates, chaplains and lay organisations that provide support to HD individuals and their families.

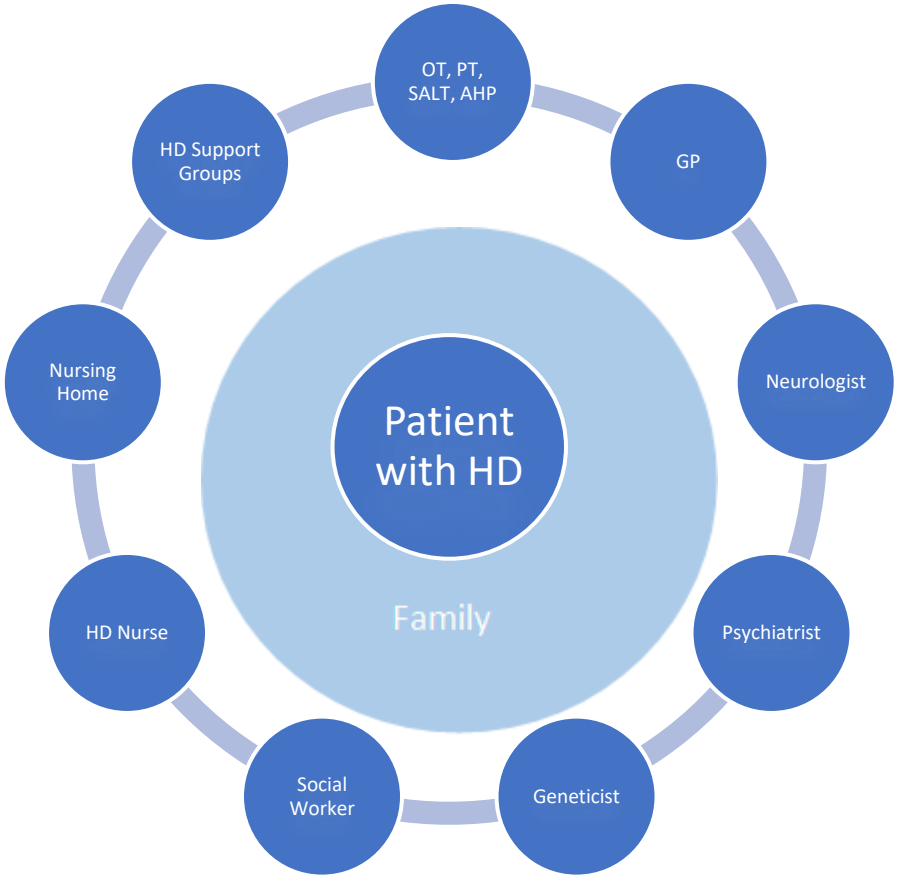


Figure 8. Multidisciplinary team (MDT) involved in HD care, adapted from Mestre and Shannon (2017)

The goal of an MDT is to alleviate HD symptoms, improve function and quality of life (Frich et al., 2016). While management of HD is centred on treating symptoms (Bates et al., 2015), the MDT must consider the overall context of the disease stage, life history, and social situation to establish a coherent treatment plan (Nance and Westphal, 2002; Nance, 2007). Coordination of HD services is central to the MDT approach. Simpson and Rae (2012) proposed an international care management pathway within HD practices to address the disparities in service delivery (Figure 9). In the UK, it has been observed that HD services are organised by clinicians who have a particular interest in the disease and that the services run in an unsystematic ad-hoc fashion (Wilson, Pollock and Aubeeluck, 2014). Wilson and Aubeeluck (2016) proposed that an HD specialist nurse can act as a key contact for patients, families and HCPs and this role can enhance care provision. Roos (2000) suggested that medical and non-medical treatment must be individually tailored, as the impact of disease differs from person to person and that the care required increases over a period of time. The HD care plan should be based on a full patient assessment within the context of their disease state, their family and the social environment.

Figure 9. The HD care pathway, EHDN Standards of Care Working Group (Simpson and Rae, 2012)

While the interventions from the MDT are widely recognised, there is a lack of research into how MDT care delivery influences outcomes in HD (Frich et al., 2016). Little has been published on HD care delivery in the community and in long-term care facilities (Edmondson and Goodman, 2017). A survey of international HD speciality clinics reported the variability of access at different HD centres and suggested that more research is needed to understand how these services are organised and how HD centres interact with local health care systems (Frich et al., 2016). Edmondson and Goodman (2017) argued the impact of MDT care in HD centres is limited as most people with HD, and their families, are not seen in speciality centres. A greater number of HD patients and their families are primarily under the care of non-specialist HCPs such as GPs (Edmondson and Goodman, 2017), district nurses or nursing home staff (Wilson, Pollock and Aubeeluck, 2014). The rarity of HD means that many community HCPs are likely to have insufficient

knowledge or understanding of HD to provide informed care. The experiences of HD families suggest that caregiving problems are associated with difficulty in accessing HD services, and the lack of support they receive from HCPs (Skirton et al, 2010, Parekh et al, 2018).

UK POLICY AND GUIDANCE

Since 2005, the Department of Health introduced the National Service Framework (NSF) for people with neurological conditions. At the core of the framework were 11 quality requirements which aim to put the individual at the heart of care, and provide a service that is efficient, supportive and appropriate from diagnosis to end of life (DH 2005). The quality requirements were fully implemented in 2015, although progress has been poor (NAO 2015). The National Audit Office (NAO, 2015) reported significant uncertainty from the start as there were no specific arrangements in place to monitor how local commissioners implement the framework in the light of changing government priorities, such as the cessation of NHS funding that decommissioned services such as Long-Term Conditions Delivery Support Team. There is evidence to suggest that care for people with neurological conditions has worsened since the NSF was published (NAO, 2015). A study by Sixsmith et. al (2013) which explored people's experiences with NSF found that there was a fragmentation of services within NSF care pathways and the services lacked financial resources and staffing capacity to implement continuous quality care. Further evaluation of the NSF reported significant problems with the care pathway, which include many patients not being given information about their condition and the support available to them, and that there was poor coordination of health and social care services (NAO, 2015). In 2012, a progress review by the Committee of Public Accounts made recommendations aimed at achieving better outcomes for people with neurological conditions, one of them being the development a neurological data set drawn from health and social care systems. This recommendation was heeded by Public Health England (PHE) and NHS England by jointly sponsoring a Neurology Intelligence Network (NAO 2015). NHS England (2016) also commissioned Thames Valley Strategic Clinical Network to lead a national clinical programme of work. In 2019, following the NAO recommendation, NICE (2019) developed quality standard guidelines for local commissioners and health care providers relating to the needs of, and services for people with neurological conditions.

Furthermore, in the wake of the Covid-19 upheaval, the NHS is undergoing significant organisational change as it attempts to move to a system of integrated care (Thomas, 2021). The service integration will adopt the core frameworks of the Long-Term Plan (NHS England 2019) and the Health and Care Bill (UK Parliament, 2021) which aims to bring services together, improve care access and coordinate care. However, the UK Neurological Alliance, a charity composed of over 70 organisations, expressed concerns over the lack of detail in the Integrated Care Structures (ICS) proposed for neurology care (Neurological Alliance 2021). Thomas (2019) observed that while some neurological conditions are given priority, other neurological diseases will 'become lost in the umbrella of long-term conditions' (p.40). UK policy and guidance primarily refers to PD and MS. There is lack of research studies on UK care provisions for HD in comparison to PD, MS and MND (Wilson et al., 2008) such that NICE Guidelines (2022) has published

quality standards for other neurological conditions except HD. It could be that the rarity of HD has resulted in the lack of UK guidelines to develop appropriate care, although MND, which has less prevalence than HD, has a NICE guideline and has achieved better awareness in research and care agendas.

Despite the scarcity of UK policy and guidance in HD, voluntary organisations such as the Huntington's Disease Association (HDA) and the European Huntington's Disease Network (EHDN) have provided a range of resources and guidance for HD treatment and care. Although these standards of care for HD individuals and their families have been published, they only serve for the purpose of service evaluation and as a foundation for further research (Frich et al., 2016).

The material in this section has tried to elucidate our understanding of HD as a rare genetic disease with a complex nature (the combination of motor, psychiatric and cognitive symptoms). Over the past centuries there has been a rapid growth in our understanding of the HD pathophysiology and clinical manifestations. These improvements, since the publication of George Huntington's seminal paper (1872) to the discovery of the HD gene (1993), have represented a massive leap forward in our knowledge, with the advent of gene-targeted therapies which have entered into human trials and the possibility of gene repair. The genetic nature of HD has been simultaneously a remarkable and a dreadful thing (Nance, 2017). A disease with a deep history rooted in witchcraft, violence and eugenics consequently has been stigmatised even more cruelly by proof of its inheritability (Wexler, Wild and Tabrizi, 2016). In addition to its genetic profile, HD is a physically, psychologically, and socially devastating familial disorder.

From the clinician's perspective, contemporary HD research has mainly focused on the biomedical aspects and genetic biomarkers (Roos, 2010; McColgan and Tabrizi, 2018; Testa and Jankovic, 2019). This section of the Literature Review has uncovered that the available HD literature in the past decade has mainly focused on genetics and the search for a cure. Despite the widespread interest in HD care models, there is a poor evidence base for current HD treatments and modes of MDT care. HD is still incurable, and while some clinical interventions can help ease the disease symptoms, the psycho-social support can significantly improve the quality of life of generations of families and individuals affected by this complex disease (Wexler, Wild and Tabrizi, 2016).

The next literature domain will look closely at HD social and relational structures, in particular the experiences of family caregivers. The subsequent sections will explore qualitative HD literature on what we know about the experiences of people living with, and suffering from HD.

THE PERSPECTIVES OF FAMILY CAREGIVERS

The SPIDER approach was used to identify the literature that facilitated understanding of the experiences of family caregivers of HD. The key terms for Sample (S) included "Family" OR "carer" OR "caregivers", and the Phenomenon of Interest (PI) included "Huntington's Disease" (D) "interviews" "focus groups",

"observations". Evaluation (E) key words included "care experiences", "care", "caregiving burden" "quality of life", "well-being" "views" and "perceptions". Research type (R) include "qualitative" and "mixed methods". Search limits were placed which included full texts studies from"2008 to present date" and published in the English language.

Most of the search was conducted in 2018-2019, a further search in March 2020 found a literature review that synthesised qualitative studies focusing on family carers' experiences impacted by HD (Parekh, Praetorius and Nordberg, 2017). The paper reviewed twelve studies published from 1993 until 2016 and utilised Qualitative Interpretative Meta-Synthesis (QIMS) and found the profound impact on the HD family systems. Parekh et al (2017) addressed the findings to social workers on how they can play an active role in improving support for the population group and recommended specific interventions such as genetic counselling and community education. In common with studies obtained from the previous literature domain on clinical perspectives of HD, the limited HD literature that explored families' and caregivers' experiences put emphasis on HD genetic aspects.

The result of the literature search has produced a number of studies on HD families' and caregivers' experiences of health care services (Soltysiak, Gardiner and Skirton, 2008; Skirton et al., 2010; Etchegary, 2011a) with more specific issues around genetic testing (Etchegary and Fowler, 2008; Hagberg, Bui and Winnberg, 2011; Smith et al., 2013). HD literature also explored families' and caregivers' views on health related quality of life (Aubeeluck and Moskowitz, 2008; Aubeeluck, Buchanan and Stupple, 2012; Carlozzi and Tulsky, 2013), communication (Hartelius et al., 2010; Zarotti, Simpson and Fletcher, 2019) and the changes brought about by the HD progression from preclinical stages (Williams et al., 2007) to palliative care (Dawson et al., 2004). Studies also explored HD family caregivers' well-being (Roscoe et al., 2009; Williams et al., 2009b; Scerri, 2015), social structures; roles and relationships (Mantell, 2010; Maxted, Simpson and Weatherhead, 2014; Røthing, Malterud and Frich, 2014). While some studies have delineated the experiences of different caregivers such as HD spouses (Lowit and van Teijlingen, 2005), children or teen caregivers (Williams et al., 2009a; Williams et al., 2013), most studies were conducted alongside the person with HD or with health professionals (Aubeeluck, Buchanan and Stupple, 2012; Carlozzi and Tulsky, 2013; Maxted, Simpson and Weatherhead, 2014).

Based on the search results, many of the qualitative studies have used semi-structured and focus group interviews. Further, there appear to be more HD quantitative studies that capture families' and caregivers' experiences. Methods driven by surveys and questionnaires obtained aspects of health-related quality of life (Ready et al., 2008; Williams et al., 2012; Carlozzi and Tulsky, 2013; Simpson et al., 2016; van Walsem et al., 2017). HD research has consistently reported the reduced quality of life in family caregivers (Aubeeluck and Moskowitz, 2008; Ready et al., 2008; Aubeeluck, Wilson and Stupple, 2011). Consequently, this recognition led to a development of the Huntington's Disease Quality of Life Battery for Carers (HDQoL-C), a validated clinical tool used to assess HD caregivers' quality of life (Aubeeluck et al., 2019).

Moreover, Parekh et al. (2017) found that there are limited empirical studies describing psychosocial experiences of HD family caregivers. The majority of HD studies, in a purely qualitative sense were sparse (Audulv, Packer and Versnel, 2014) and the majority were conducted outside the UK (Williams et al., 2009b; Røthing, Malterud and Frich, 2014; Scerri, 2015). This thesis chapter could argue that the difference between health care settings and other cultural factors would largely affect the outcome of these studies. For example, Williams et al (2012) examined the personal concerns of HD family caregivers in both the US and the UK and found that notable issues vary with US respondents highlighting the financial constraints and impact on children, while in the UK respondents emphasise emotions and sadness. While the nature of qualitative research is not generalizable and individuals ascribe personal meaning to their experiences, the literature has however highlighted the wide impact of HD on family caregivers. The following literature section explores the family and caregivers' experiences. Moreover, a section of this literature domain will summarise the similarities and differences of HD care with other chronic and progressive neurological diseases.

THE HD FAMILY

There is space here to discuss the earlier research that explored the experiences of HD families and caregivers. The recognition that HD care has improved over the past two decades (Testa and Jankovic, 2019) supported the inclusion criteria of this thesis that mainly focused on studies conducted within the last 10 years. However, it is important to revisit the earlier experiences of HD families and caregivers to give context on the HD experience and how it has been shaped over the course of time.

Wexler (2008) pointed out that the challenges faced by HD families is deeply rooted in the long history of prejudice and misunderstanding that have contributed to the shame and stigma of the disease. Wexler (2010) further argued that, although the eugenic origins and the harmful psycho-social legacies of HD families cannot be undone, the research and scientific community can advocate a better understanding of HD as this can influence better future care of HD individuals and families. Soltysiak et al (2008) pointed out that there is limited research on the non-clinical management of the illness experienced by affected individuals and families. A large survey of 3,600 respondents found that both the person with HD and their caregivers are severely impacted by the cognitive and behavioural symptoms of the disease (Simpson et al., 2016). Dawson et al (2004) reported that people with HD have crucial life changes imposed on them such as loss of job, mobility, and independence, in addition to the complex cognitive and psychiatric changes such as aggression and depression. The experiences of living with HD extend beyond the affliction of the HD individual to the insufferable impact on families and caregivers (Dawson et al., 2004; Edmondson and Goodman, 2017). This reinforces what Ranen et al (1995) found that suffering brought about by HD to the individual and their caregiver is mutually dependent, and that better care for the person with HD was linked to their caregivers' better ways of coping.

It could be argued that the needs of HD family caregivers are often overlooked, and researchers referred to the HD families as 'invisible patients' (Domaradzki 2015 p. 932), the spousal caregiver is the forgotten

person in the HD family (Kessler, 1993) and the HD children are 'living in the shadow' (Sparbel et al.,2008, p.327). Similarly, more recent interview studies on HD families describe the presence of HD as 'A spectre hanging over' (Maxted et al., 2014, p.339), and their caregiving experiences as 'under a cloud' (Mantell, 2010, p.33). HD places a considerable psychological and emotional burden on families (Williams et al., 2009b; Aubeeluck, Buchanan and Stupple, 2012). Studies by Dale et al (2014) and Parekh et al (2017) proposed that the specific challenge for HD caregivers is linked with the hereditary nature of the disease. Researchers widely recognised that family caregiving does not end with the death of the affected family member but repeats in successive generations. Tyler (1996) observed that HD families experience a great deal of grieving and have repeated bereavements to endure. HD is a family disease that affects more than one member of the family (McGarva, 2001), consequently a family caregiver may care for many HD sufferers in more than one generation (Kessler, 1993; Lowit and van Teijlingen, 2005).

Earlier studies reported the neglected needs of the HD individuals and their family caregivers (Kessler, 1993; McGarva, 2001; Lowit and van Teijlingen, 2005; Aubeeluck and Buchanan, 2006; Pickett Jr, Altmaier and Paulsen, 2007). While research has recognised the unique caregiving burden of HD (Pickett Jr, Altmaier and Paulsen, 2007), some studies also highlight the negative coping strategies of avoidance (Lowit and van Teijlingen, 2005), guilt of passing HD to children (Kessler, 1993), and the survivor guilt of other family caregivers who obtained a negative test (Dawson et al., 2004). Of all the difficulties experienced by HD family caregivers, interview HD studies have consistently reported that the major burden of HD caregiving is attributed to their interaction with health professionals and heath care services (Kessler, 1993; McGarva, 2001).

The succeeding sections will present the literature over the past decade on HD family caregivers to determine if there has been much change in their experiences over time and the review has so far identified the predominant themes which include their interactions with health care services, the changes in the family structures, the caregivers' burden of care and their coping strategies (Figure 10).

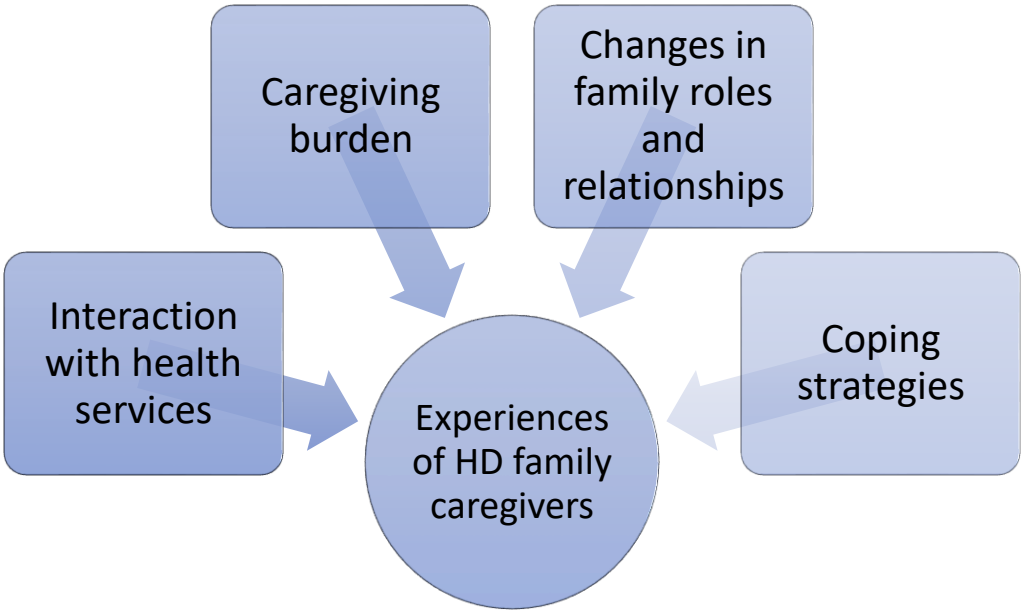


Figure 10. **The four broad areas of experiences of HD family caregivers**

EXPERIENCES WITH HEALTH CARE

The available HD literature has explored the varied experiences of family caregivers' interaction with health and social care services. Edmonson and Goodman (2017) attributed the wide disparities of HD care experiences and stated that contributing factors include the varying access to HD services, and other attitudinal and structural barriers. Attitudinal barriers have prevented people from engaging with care services due to negative assumptions and past experiences (Edmondson and Goodman, 2017). Other factors include genetic discrimination (Bombard et al., 2008; Williams et al., 2010), stigma (Wexler, 2010) and denial (McCusker and Loy, 2014). Structural barriers are often imposed by health care systems which include geographic distance (Edmondson and Goodman, 2017), medical care costs (Williams et al., 2012; Jones et al., 2016), lack of, or limited access to, specialised care (Aubeeluck, Buchanan and Stupple, 2012; Wilson, Pollock and Aubeeluck, 2014; van Walsem et al., 2017) and the physical disability of the person with HD (Soltysiak, Gardiner and Skirton, 2008) more so in later stages (Edmondson and Goodman, 2017). Similarly, Scerri (2015) identified that the 'micro context' (relating to the individual) and the 'macro context' (concerning the societal, national, and international/global level) have contextual impact on families and their caregiving duties.

The HD family caregivers persistently report their dissatisfaction and frustration with health care services (Soltysiak, Gardiner and Skirton, 2008; Etchegary, 2011; Aubeeluck, Buchanan and Stupple, 2012; Scerri, 2015). Interviews with family caregivers in the Canada found that many physicians have no real understanding of HD (Etchegary, 2011a). A sentiment shared by families in the UK on the lack of HD practical knowledge among health professionals (Skirton et al., 2010; Aubeeluck, Buchanan and Stupple, 2012). A few studies investigated the general health care experiences of HD affected families ,caregivers and individuals in the UK (Aubeeluck, Buchanan and Stupple, 2012), Canada (Etchegary, 2011a), Poland

(Domaradzki, 2016), Norway (Røthing, Malterud and Frich, 2015b), and the USA (Roscoe et al., 2009; Williams et al., 2009b). Moreover, some studies explored specific HD care provisions in a community setting (Soltysiak, Gardiner and Skirton, 2008), specialist inpatient service (Dale, Freire-Patino and Matthews, 2014) and rehabilitation units (Frich, Røthing and Berge, 2014). Experiences of genetic testing comprise the majority of the literature on experiences of HD family members (Etchegary and Fowler, 2008; Smith et al., 2013). A systematic review by Crozier et al (2015) acknowledged the notable psychological impact of genetic testing that disrupts the family systems and suggest that appropriate testing protocols to support individuals and families warrant further research.

In addition, HD family caregivers acknowledge the importance of dedicated HD services (Soltysiak, Gardiner and Skirton, 2008; Kenny and Wilson, 2012) and the valuable role of a knowledgeable HD professionals such as a specialist nurse (Wilson and Aubeeluck, 2016) and expert clinicians (Edmondson and Goodman, 2017). However, these specialist inputs are not readily available or only provided in circumstances of crisis (Dawson et al., 2004; Soltysiak, Gardiner and Skirton, 2008). A questionnaire study of 55 family caregivers found that their perception of caregiving was hugely influenced by their negative experiences with the health care system (Domaradzki, 2016). The lack of HD knowledge to provide informed care by primary care and community services continues to be a significant challenge to HD family caregivers, who are often without access to specialist care (Edmondson and Goodman, 2017).

Soltysiak et al (2008) recommend that available HD services need to be flexible in approach, more so with the psychological and practical support they provide. While there is recognition of the need for more comprehensive resources and care facilities in HD care, HCPs and policy planners also need to be aware of the wider needs of HD family caregivers (Domaradzki, 2016). Inclusive speciality outreach to HD-affected families in their home community and better training and support to community providers is a feasible approach to improve HD care (Edmondson and Goodman, 2017). In addition, acknowledging the competent roles and contribution of HD family caregivers can help partnership working and promote continuity of contact throughout the care course (Aubeeluck and Moskowitz, 2008; Røthing, Malterud and Frich, 2015b; Edmondson and Goodman, 2017) which can help improve coordination of HD care.

THE BURDEN OF CARE

Another common theme found in this literature review is the burden attributed to the caregiving roles of HD family members. Care for people with HD is usually provided by family members, who themselves are at risk of HD or have children who are at risk of inheriting the disease (Dale, Freire-Patino and Matthews, 2014; Aubeeluck et al., 2019). Families can be overwhelmed by the thought of providing care for siblings and other relatives (Roscoe et al., 2009). Domaradski (2016) found that families are burdened by the amount of time they spend in caregiving and the magnitude of the tasks involved. The hereditary nature of HD means that family members may be simultaneously afflicted, which further increases the amount of the caregivers' load. Family members could often find themselves in long term caregiving roles that supersede

their own life choices (Aubeeluck, Buchanan and Stupple, 2012), Many family members neglect their own needs as their caregiving role tends take to over their own personal lives (Aubeeluck and Moskowitz, 2008). This theme is consistent with interview studies that found that HD family caregivers experience the disintegration of their own lives (Williams et al., 2009b) and that their family network becomes vulnerable and fragmented (Røthing, Malterud and Frich, 2014).

The financial burden of the disease on families is revealed in a few qualitative studies (Williams et al., 2012; Røthing, Malterud and Frich, 2014). Jones et al (2016) argued that the societal cost of HD is an underestimated burden. A UK study found that HD care provided by family caregivers (informal care) is the largest driver of care costs in all HD stages and the needs of their caregivers must be included in care planning (Jones et al., 2016). A review by Edmonson and Goodman (2017) found that most individuals with HD and their families in the US, UK and Australia are not seen in HD speciality centres, regardless of the differences in health care systems. Similarly, a European survey found the large reliance on informal HD care which is usually provided at home and to a lesser extent in hospital-based services (Busse et al., 2011). Researchers recommend that the needs of family caregivers must be taken into account in HD care provisions (Busse et al., 2011; Divino et al., 2013; Jones et al., 2016) as the practical and emotional burden of caring for HD significantly impacts quality of life (Ready et al., 2008; Williams et al., 2009b; Aubeeluck, Wilson and Stupple, 2011; Aubeeluck, Buchanan and Stupple, 2012; Aubeeluck et al., 2019).

The recognition of the HD burden of care allowed researchers to focus on the caregivers' quality of life (QOL) (Aubeeluck, Wilson and Stupple, 2011; Aubeeluck, Buchanan and Stupple, 2012; Aubeeluck et al., 2019). Roscoe et al (2009) found that despite the high level of perceived stress in HD family caregivers, the participants also reported relatively high levels of life satisfaction. However, the study further cautioned that the HD families were recruited from HD Centres of Excellence in medical care (Roscoe et al., 2009), and not all families have access to such specialist services (Edmondson and Goodman, 2017). Long term HD caregivers are potentially placing their physical and mental health at risk (Roscoe et al., 2009). The HD caregiving burden increases over the length of time imposed by symptoms progression (Novak and Tabrizi, 2011) , caregiving tasks (Domaradzki, 2016) can become too heavy, increasing the risk of poor care at home (Roscoe et al., 2009). Family caregivers eventually resort to respite care facilities which often leads to care transitions and institutionalised 24-hour care (Simpson, 2007; Roos, 2010; Novak and Tabrizi, 2011). Roos (2010) recommended that throughout the HD caregiving process, psychological support needs to be offered to family caregivers as their cumulative responsibilities can considerably impact their relationships with afflicted HD family members.

CHANGES WITH ROLES AND LOSS OF RELATIONSHIPS

Interviews with family caregivers have reported the end of spousal relationships or parent-child relationships being replaced by relationships bounded by duty of care (Sparbel et al., 2008; Williams et al., 2009b; Mantell, 2010; Røthing, Malterud and Frich, 2014). Williams et al (2009) found that spousal caregivers adapted to the situation by 'falling out of love' to resolve marital conflicts and to allow

themselves to provide care. Aubeeluck et al (2012) found that one of the most distressing issues applicable to spousal caregivers was a sense of loss in marital relationships. Similarly, Rothing et al (2014) noted that reciprocal relationships have been difficult to maintain, these transitions of relationships occurred gradually as the need for care increased. The spousal caregivers reported the loss of relationship and their hopes for the future (Mantell, 2010). HD spouses expressed that they have replaced their romantic love with caregiving, similar to losing an equal partner and instead bringing a new child to look after (Williams et al., 2009b; Røthing, Malterud and Frich, 2014). A study of several women providing personal intimate care to their husbands with HD compared their experiences to caring for a young child, and that they were unable to view their partners sexually (Mantell, 2010). In the same way that children of affected parents shift their roles from child to parental caregivers, a role reversal process which Maxted et al (2014) referred to as parentification, the hereditary nature of the HD has led families to negotiate their reciprocal roles (Maxted, Simpson and Weatherhead, 2014). Consistent themes shared by studies on HD children caring for their HD-affected parents found that while family members move into different roles according to their position in the family, they are also confronted with the worry of their own future (Sparbel et al., 2008; Williams et al., 2009a; Maxted, Simpson and Weatherhead, 2014).

A comparative study of US and UK HD family caregivers found that the strain of long-term care responsibility was brought about by the HD psychiatric symptoms and complex motor and cognitive function (Williams et al., 2012). HD progression significantly impacts the family roles leading to loss of relationships (Mantell, 2010; Williams et al., 2012). Williams et al (2012) observed the loss of meaningful relationships between family caregivers, and other studies have noted their feelings of being misunderstood (Aubeeluck, Buchanan and Stupple, 2012), worries of maintaining family goals and financial stability (Mantell, 2010; Røthing, Malterud and Frich, 2014), and the loss of affection between children and their affected parents or other family members with HD (Sparbel et al., 2008; Mantell, 2010; Williams et al., 2013). Maxted et al. (2014) highlighted the complex relationships in HD families, causing them to come together and apart, with the constant changes and adaptions to new roles.

COPING STRATEGIES

Interviews with family caregivers have revealed different coping strategies relating to their experiences of caring for people with HD. In a study of forty-two caregivers, Williams et al (2009) found caregivers attempt to cope by seeking comfort from other family members, using prescribed medications, and anticipating the time of death of the care recipient. HD caregivers experience a sense of ongoing loss, and anticipatory loss has been central to caregivers' experiences, this is inherent to the hereditary component of the illness (Sobel and Cowan, 2003; Vamos et al., 2007; Mantell, 2010). A UK focus group study of forty seven family members noted a range of negative emotions including guilt, anger, depression and frustration combined with a sense of loss in their caregiving roles (Aubeeluck, Buchanan and Stupple, 2012). Coping with the sense of loss was not exclusive to HD family caregivers, Carreon et al (2018) also found that difficulty in coping is present in nurses and health care assistants in HD long term care facilities. The findings of the study, conducted in a UK nursing home, suggest that care providers should provide

emotional support to families and support caregivers' coping strategies to help improve HD care (Carreon, Hayes and Leavey, 2018). Studies by Soltysiak et al (2008) and Scerri (2015) similarly recommended that health policies and HCPs can assist with HD family caregivers' psychological wellbeing by facilitating the development of better coping strategies.

HD family caregivers have used a diverse range of coping strategies including religion and spirituality (Roscoe et al., 2009; Røthing, Malterud and Frich, 2014), sharing experiences with HD support groups (Soltysiak, Gardiner and Skirton, 2008; Dale, Freire-Patino and Matthews, 2014), finding meaning and appreciation of the positive aspects of life (Roscoe et al., 2009; Mantell, 2010; Røthing, Malterud and Frich, 2014). Roscoe et al (2009) referred to these examples as protective factors that mediate the stresses associated with HD caregiving. Etchegary (2009) however revealed that while these coping strategies are varied and dynamic, they could be classified into three categories, the primary, secondary and social comparison strategies. Despite the study samples at different stages of HD, the commonality between these two studies (Etchegary, 2009; Roscoe et al., 2009), found that participants actively sought social support networks with other HD caregivers. HD caregivers valued spending time with other caregivers as this can alleviate loneliness and provide the opportunity to talk and listen to similar HD care experiences (Dale, Freire-Patino and Matthews, 2014). The feeling of being not able to belong is common (Aubeeluck, Buchanan and Stupple, 2012), family caregivers often feel they are not being understood by others (Roscoe et al., 2009; Williams et al., 2009b). Therefore, by sharing their caregiving experience, in return they have avoided the negative social consequences and stigmatisation of the disease (Røthing, Malterud and Frich, 2015a). This kind of support differs from practical support, as some caregivers state the benefit of social support groups has been a source of their strength (Aubeeluck, Buchanan and Stupple, 2012).

Avoidance has been a prominent theme in family caregivers' experiences (Parekh, Praetorius and Nordberg, 2017). This is not surprising as it is revealed in a study by Etchegary (2009) that acceptance is the least frequent strategy that caregivers used. A small study of ten spousal caregivers observed that many denied the HD symptoms of their partners, pretended that HD progression was not happening and consequently delayed seeking care until the symptoms became prominent (Lowit and van Teijlingen, 2005). Avoidance has likewise proved to be an impediment in communication (Hartelius et al., 2010) and caused negative long term consequences (Etchegary, 2009). A study on family dyads noted that family caregivers tend to avoid talking about the illness both with the family units and outside their circle (Maxted, Simpson and Weatherhead, 2014). While this avoidance strategy has provided some relief to deal with anxiety, however it has hindered family caregivers to seek appropriate care interventions such as making anticipatory plans for the future (Lowit and van Teijlingen, 2005; Etchegary, 2009).

The available literature has provided insight into the numerous coping strategies that HD family caregivers employ, and this in the context of their daily lives and family members' risk of fatal disease. The different ways that HD families and caregivers adapt to change has been presented in various studies (Williams et al., 2009b; Røthing, Malterud and Frich, 2015a; Scerri, 2015). Williams et al (2009) noted that HD caregivers

adapt to changes as they happen and use an operational rather than strategic approach to care. Other studies reinforced this observation that family caregivers deal with and adjust to problems as they come (Maxted, Simpson and Weatherhead, 2014). While the impact of illness has taken a toll on the emotional experiences of families and caregivers (Williams et al., 2009b; Carlozzi and Tulsky, 2013), the presence of HD is rarely talked about in the family (Etchegary, 2009; Maxted, Simpson and Weatherhead, 2014).

HD CARE IN COMPARSION WITH OTHER CHRONIC AND PROGRESSIVE NEUROLOGICAL DISEASES

The experiences of HD family caregivers appear to be centred on interaction with health care services as is the case in other chronic and neurological conditions. Parkinson's Disease (PD), Multiple Sclerosis (MS), Motor Neuron Diseases (MND), and other types of Brain Injuries and Dementias share the most obvious feature of progressiveness, which means the symptoms become more severe and the disability tends to increase over time. These diseases are collectively referred to as progressive long-term neurological conditions (PLTNC). People affected with PLTNC need further care interventions and support over time (Venning et al., 2008; Methley et al., 2015). Wilson et al (2008) conducted a literature review on PLTNC to determine areas of best practice which could promote improvements in UK health services, and the study suggested it is vital to maintain support and contact with family caregivers to deliver timely care. Similarly, Whitehead et al (2018) explored the role of family caregivers in other chronic diseases such as COPD and Diabetes and found that it had a strong influence on health and illness outcomes. Health providers play a vital role in assessment, offer support, and explore coping strategies of families and affected individuals (Wilson et al., 2008; Peters et al., 2013; Whitehead et al., 2018). However, researchers argue that barriers for optimal care continue to exist across long term conditions (Bridges, Flatley and Meyer, 2010; Jeon et al., 2010; Chaplin, Hazan and Wilson, 2012; Ploeg et al., 2019) and these include the lack of specialist knowledge on neurological conditions (Wilson et al., 2008; Gallacher et al., 2013), the difficulty of accessing specialist care (Chaplin, Hazan and Wilson, 2012; Methley et al., 2015) and the lack of awareness on the needs and care of individuals and their families (Bridges, Flatley and Meyer, 2010; Ambrosio et al., 2015; Vescovelli, Sarti and Ruini, 2018).

Specialist support provided by the MDT and specialist centres is widely promoted in the care of chronic conditions but often these services are fragmented and in some areas are not accessible (Frich et al., 2016; Harris et al., 2018). This is a consistent message in HD literature and evident in other progressive long-term conditions, that families and caregivers are critical of the lack of support and health care services available to them (Fitzpatrick et al., 2010; O'Connor and McCabe, 2011; Peters et al., 2013). Skirton et al (2010) reported that the insufficient knowledge in community-based health services on HD care is a major concern of HD families and caregivers in the US and the UK. The lack of coordination of health services is widely recognised in the literature (Gallacher et al., 2013; Ploeg et al., 2019), despite research studies pointing out that delaying treatment contributes to worsening symptoms, which in turn makes a significant contribution to caregivers' burden (Aubeeluck, Buchanan and Stupple, 2012; Bergin and Mockford, 2016)

and has a negative impact on the quality of life of individuals and their families (Fitzpatrick et al., 2010; Peters et al., 2013; Ploeg et al., 2019).

O'Connor and McCabe (2011) reiterated the critical role of PLTNC family caregivers with the shift of health care systems from institutional to community care and peoples' preference to be cared for at home. Equally, Peters et al (2013) recognised their fundamental role of complementing the support provided by health and social care services. A cross-sectional study of PLTNC caregivers found they have unmet needs and often have less access to services to help them (Peters et al., 2013). Therefore, it is important to improve caregivers' quality of life where possible, as the impacts on the caregiver and the care recipient are considerable (O'Connor and McCabe, 2011). Qualitative interviews on family dyads of MS (Golla et al., 2015) and HD (Maxted, Simpson and Weatherhead, 2014) report the unmet needs, as caregivers tend to neglect their own personal wishes. Moreover, research on families of PTLNC and other chronic diseases highlight general experiences of caregivers' reduced well-being (O'Connor and McCabe, 2011; Mosley, Moodie and Dissanayaka, 2017; Harris et al., 2018), notably found in spouses (O'Connor, McCabe and Firth, 2008; Hawkley et al., 2020), and children caregivers (Pinquart and Sörensen, 2011). Families further revealed aspects of the caregiving burden (Mosley, Moodie and Dissanayaka, 2017; Whitehead et al., 2018; Sheehan et al., 2019), including time and financial burdens (O'Connor and McCabe, 2011; Mosley, Moodie and Dissanayaka, 2017; Sheehan et al., 2019), coping strategies (Mosley, Moodie and Dissanayaka, 2017) and describe the changes and loss of relationships brought about by caregiving duties (O'Connor, McCabe and Firth, 2008; Pinquart and Sörensen, 2011; Bunn et al., 2012; Whitehead et al., 2018).

THE UNIQUENESS OF HD

This literature section has uncovered that HD caregivers share comparable experiences with caregivers of other progressive and chronic conditions. The findings of this review further support the limited literature on HD family caregivers in comparison to other PLTNC (Wilson et al., 2008), and that although they have similar experiences to other family caregivers, caring for HD is certainly unique (Williams et al., 2009b; Domaradzki, 2016; Parekh, Praetorius and Nordberg, 2017).

Firstly, HD care is distinguished from other neurological diseases by its slower and longer disease progression that could span up to three decades. The disease duration strains the ability of families to provide care over many generations (Soltysiak, Gardiner and Skirton, 2008; Sparbel et al., 2008; Williams et al., 2009a; Mantell, 2010). Additionally, the genetic transmission is an enduring worry for HD family caregivers that they too can have the risk of developing the disease. There is also the feeling of guilt by HD spousal caregivers of passing the illness to their children (Williams et al., 2009a; Williams et al., 2009b). The emotional burden of HD families is that they have to witness multiple immediate and extended relatives dying from the illness.

Secondly, HD symptoms generally manifest in middle age, and consequently their primary caregivers are immediate families (Aubeeluck, Buchanan and Stupple, 2012), usually their spouses who are also younger

and often tending to have children and professional responsibilities (Williams et al., 2009b). HD caregivers are relatively young compared to caregivers of PD (Mosley, Moodie and Dissanayaka, 2017) and Alzheimer's Dementia (Lethin et al., 2017). As HD progresses and caregiving becomes more demanding, family caregivers often ignore their own needs (Røthing, Malterud and Frich, 2014) and abandon their leisure activities and professional careers (Domaradzki, 2016). Focused family interventions such as problem solving (Jona et al., 2017)and psycho-emotional support (Williams et al., 2009b) are often needed by family caregivers who have to deal with increasing responsibilities while losing contact with their partners deteriorating from HD (Roos, 2010).

Thirdly, the complex degenerative course of HD requires more intensive care for individuals and their family caregivers. The burden of caring at home can become too heavy and people with HD in their later stages would require additional help at home (Roos, 2010), or 24-hour full nursing care (Aubeeluck, Buchanan and Stupple, 2012). Although earlier studies have reported that the existing aged care facilities (residential, respite and nursing homes) are clearly unsuitable to meet the needs of the younger patients with HD (Dawson et al., 2004; Simpson, 2007), Edmondson and Goodman (2017) reported that HD long term care facilities are inaccessible to the vast majority of HD individuals regardless of cost or proximity. The lack of access to specialist HD care is a common theme in multiple studies relating to the burden experienced by family caregivers (Skirton et al., 2010; Aubeeluck, Buchanan and Stupple, 2012).

Lastly, HD is a rare disease of which community health and social care providers have limited understanding, not only on the clinical and practical needs of HD patients but also the disease burden it poses to families and caregivers (Aubeeluck, Buchanan and Stupple, 2012). The general unawareness of HD in society makes it difficult for family caregivers to access appropriate support (Mantell, 2010). The limited availability of community support makes the experiences of HD family caregivers different from other family caregivers of other more common progressive conditions such as PD, MS and other diseases associated with ageing.

The literature review that focuses on family caregivers has uncovered the profound impact of HD that reaches far beyond the suffering of the individual to affect generations of families. The findings of this literature domain further draw attention to the mutual consequence of the disease influenced by the multi-faceted needs of the HD family caregivers and their care recipients. The available literature on the experiences of family caregivers has highlighted their caregiving burdens and perceived lack of support available to them.

Based on the findings of this review, despite the developments in therapeutics and clinical HD care, there appears to have been limited improvements according to the voices of HD families and caregivers. HD literature in the 2000s, particularly in the last decade echoed the same sentiments and burdens of family caregivers in the 1990s. Although some personal concerns vary amongst studies and across national boundaries, there are common themes related to the caregiving burden of HD families. The lack of basic HD knowledge both in society and health care professionals and accessing appropriate healthcare are the

significant challenges reported by family caregivers despite the differences in geographical boundaries, care provisions and cost. Moreover, contemporary care providers remain unaware of the ongoing health needs of HD individuals, and even more so with the needs of their family caregivers. Edmondson and Goodman (2017) poignantly described that the HD individual and their family caregivers face; 'considerable suffering, destructive disease behaviours, interpersonal tension, decades of loss and many opportunities for transformation together' (p.167).

The widespread literature suggests that although HD family caregivers share similar concerns to those of family caregivers of other progressive and chronic diseases, HD care is considerably distinct. This is particularly due to its rarity, genetic transmission, younger disease onset, longer disease trajectory and the exhausting effect on family systems. HD stands out as one of the most devastating diseases, not only because of its progressive neurodegeneration, but also its compounding impact on families and generations. The findings of this review do suggest that could there is substantial evidence of the problems and needs of HD family caregivers reported in the literature, with the growing research interest in family caregivers' quality of life (QoL). Moreover, there are available clinical tools to assess the caregivers' QoL (Mestre et al., 2018; Aubeeluck et al., 2019). However, similar to the findings of the clinical perspectives' domain of this review chapter, the literature has focused on the hereditary consequences of HD and very little is known of the effective interventions that ameliorate the sources of the family caregivers' burden.

This literature review has so far looked at the experiences of HD from the perspectives of clinicians and family caregivers, which leads us to examine the individual perspective in the final domain of this thesis chapter. It will explore the experiences of individuals living with a disease that Huntington's bestowed on them.

THE INDIVIDUAL PERSPECTIVE

The third domain, which is the final part of this chapter presents the literature on the illness experience of individuals with HD. The first domain has been driven by HD clinicians' discourse on the biomedical aspect of the disease. Based on the findings of this review, emphasis has been put on the management of HD symptoms. The second domain has explored the personal experiences of HD family caregivers. The review highlighted the pervasive impact of HD on family systems and relationships. Moreover, the two domains (the perspectives of clinicians and family caregivers) suggested that genetic concerns constitute the predominant topic in HD literature.

This final domain will look at HD from the perspectives of HD individuals on how they view their illness and the bearing of their illness experience. This section will begin with an overview of illness experience followed on by a scoping review of the HD personal experience. Finally, this chapter will present a summary of the perspectives of clinicians, family caregivers and HD individuals. These encompassing perspectives will facilitate our understanding of the holistic experiences of people living with and affected by HD as identified in current literature. This understanding will help examine the illness experience of HD

which will be further explored by this thesis. By identifying the gaps in HD literature this will help inform the research inquiry and will aid in the development of the research design and methods of data collection.

THE ILLNESS EXPERIENCE OF THE PERSON WITH HD

What we know from studies on the individual experience of HD and particularly distress around diagnosis is hugely influenced by people's past experiences (Leontini, 2006; Schwartz, 2010). These include the implications of the disease on the individual, and what they have experienced and witnessed with parents or other family members (Helder et al., 2002; Halpin, 2018). A cross-sectional survey of 77 individuals with HD found that disease perception was characterised by a strong illness identity, compounded by negative beliefs about the long disease progression with no cure or improvement (Helder et al., 2002). Similarly, Simpson et al (2016) found that individuals with HD are most concerned about their inability to maintain independence. Arran et al (2014) suggest that people with HD have lost their sense of control of their treatment and the disease progression and other studies have reported that thoughts on end of life are common concerns (Booij et al., 2014; Carlozzi et al., 2016). This gradual degeneration of HD has a profound impact on the individuals' quality of life (Mestre et al., 2018). Research has suggested that as much as the disease symptoms severely impact the person's physical health, it is the psychological well-being that is tremendously affected (Helder et al., 2001). In addition to the devastating effects of the disease symptoms, other studies have detailed the experiences of disease stigma and discrimination (Bombard et al., 2007; Williams et al., 2010), coping with the genetic risks (Etchegary and Fowler, 2008; Etchegary, 2009), the consequences of the HD diagnosis on their families (Smith et al., 2013; Jona et al., 2017) and impacts on physical and psychosocial well-being (Ho, Hocaoglu and European Huntington's Disease Network Quality of Life Working, 2011).

Although HD qualitative studies have provided us insight with specific issues of living with HD, there appears to be no systematic aggregation of these studies that have focused on the HD illness experience. The synthesis of literature in this domain can provide a more comprehensive and deep understanding of the contextual dimensions of illness experience (Walsh and Downe, 2005). This body of knowledge has been found useful in understanding the experiences of other well-studied neurological conditions such as dementia (Harman and Clare, 2006; Górska, Forsyth and Maciver, 2018), stroke (Salter et al., 2008) and other brain disorders (Hartley et al 2014). Equally, studies have methodically explored qualitative experiences of chronic diseases (Thorne et al., 2002), chronic pain (Crowe et al., 2017), mental illnesses (Kaite et al., 2015; Walsh et al., 2016) and more common progressive long-term neurological conditions (PLTNC) such as Parkinson's disease (Soundy, Stubbs and Roskell, 2014; Vescovelli, Sarti and Ruini, 2018) and Multiple Sclerosis (Wilkinson and Nair, 2013; Parker et al., 2020).

Despite all these developments in HD research and other neurological diseases in general, Halpin (2018) argued that the sociological aspect of HD receives very little attention despite the illness causing comprehensive suffering on a societal and individual level. The majority of research on people with HD has focused on assessing changes through clinical tests and laboratory settings (Harding, Stewart and Knight,

2012). Moreover, Audulv et al (2014) conducted a literature review concerning life with a neurological condition in response to an earlier study by Thorne et al (2002) which identified gaps in our knowledge and direction of qualitative research in chronic illnesses. The study argued that although research on the experience of living with a chronic neurological condition has dramatically increased over the past decade, conditions that affect motor and cognitive abilities such as HD were to an extent greatly neglected (Audluv, Packer and Versnel, 2014). This section aims to review the current literature on the HD experience and will give insight into what we have learned so far from the HD experience, coming from the important individual perspective.

A SCOPING REVIEW OF HD ILLNESS EXPERIENCE

A scoping review provides a rigorous and transparent method that is increasingly used in research topics that have not been extensively reviewed or which are regarded as multifaceted and complex in nature (Pham et al., 2014; Peters et al., 2015). This systematic method is useful in HD research given the paucity of qualitative literature on the HD lived experiences (Mahmood, Law and Bombard, 2021) and dearth of studies that examines the impact of illness on the everyday lives of individuals with HD (Audulv, Packer and Versnel, 2014).

However, this study deviated from the standard scoping protocol (Arksey and O'Malley, 2005) given that it solely focused on qualitative studies, and the searching and screening of the relevant studies were only conducted by one reviewer after obtaining guidance from supervisors who consequently provided an oversight of the scoping review. Moreover, the Critical Skills Assessment Programme (CASP) although not generally used in scoping reviews (Pham et al., 2014) has been used as an academic exercise to appraise the quality and overall rigour of the included studies. However uncommon, the CASP assessment tool and exclusively synthesising qualitative studies have been used in other nursing and public health-related scoping reviews (O'Donovan et al., 2019; Bartlett et al., 2020; Nyanyiwa, Peters and Murphy, 2022) and have therefore informed the decisions and conduct of this review.

Consequently, the methodological framework was adopted from Arksey and O'Malley (2005) which included the following steps, (1) Identify the research question 'What is known from existing literature on the illness experiences of people living with HD?', (2) Identify relevant studies, (3) Report study selection, (4) Chart the data and (5) collate, summarize, and report the results. The optional 'consultation stage' of the framework was not conducted in this review (Arksey and O'Malley, 2005). The five stages of the framework process are outlined in Figure 11.

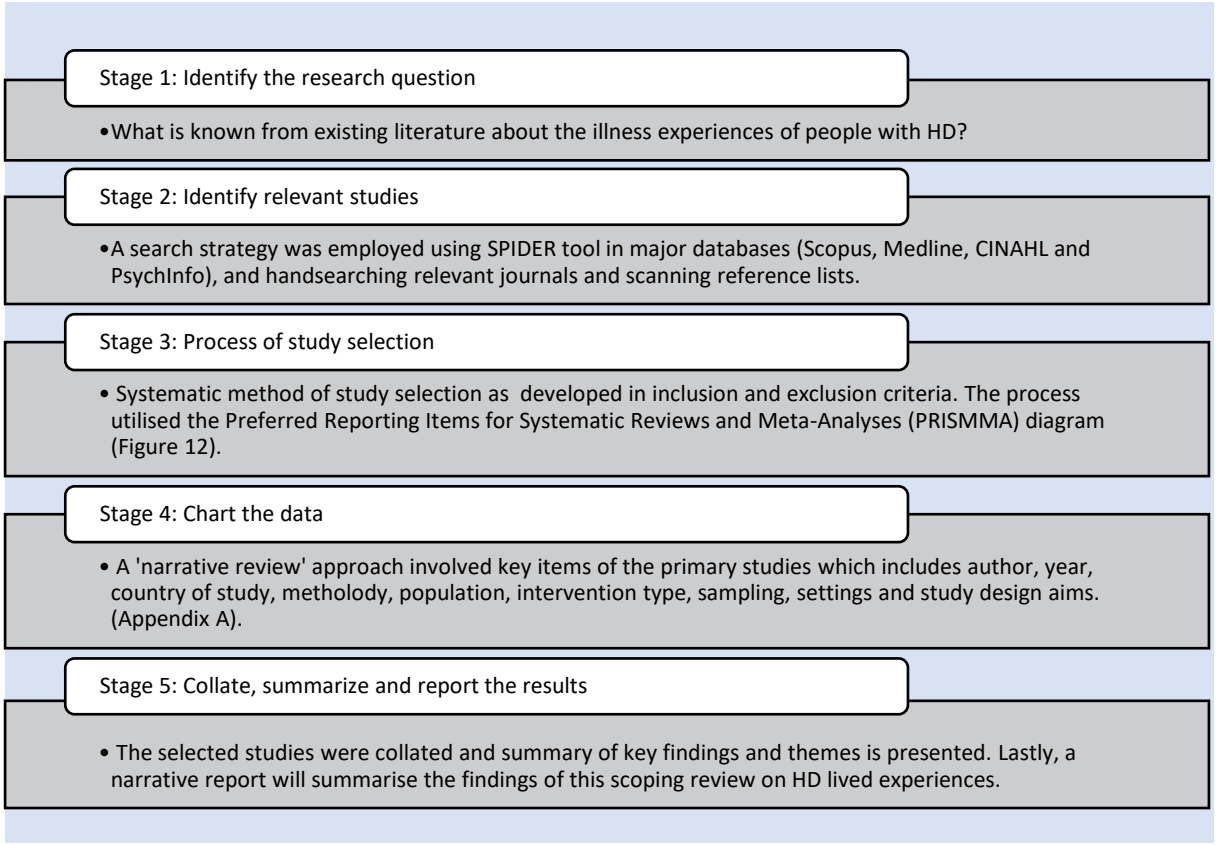


Figure 11. The five stages of the scoping framework adapted from Arksey and O' Malley (2005)

SEARCH STRATEGY

Preliminary search terms were framed applying the search strategy SPIDER (Cooke, Smith and Booth, 2012). In comparison to other search tools used in synthesising qualitative and mixed method studies such as PICOD and PEO, the components of SPIDER easily correspond to the elements of the search question and search criteria (Methley et al., 2014). The key terms for Sample (S) include "Patient*" OR "adult*" OR "person*", and the Phenomenon of Interest (PI) included "Huntington's Disease" (D) "interviews" "focus groups", "observations", "lived experience"," narratives", "case studies". The Evaluation (E) key words included "experiences", "lived", "perspectives" "quality of life", "well-being" "views" and "perceptions". Research type (R) included "qualitative" and "mixed methods". The search limits included full text studies in the English language, and to ensure the inclusion of only relatively current HD qualitative research of the past ten years (Sandelowski and Barroso, 2006) from "2018 to present". An example of the search query adapted from each academic database can be seen in Table 3, Appendix 3.

The main keywords included Huntington's disease (HD) and illness experiences. Terms for qualitative methodology include phenomenology, grounded theory, ethnography, case studies and narratives. The phenomenon of interest focused on the lived experience of HD, therefore studies that explore specific interventions, care treatments or a specific analysis were excluded. The search inclusion and exclusion

criteria are listed in Table 1, Appendix A. The literature search was performed from October to November 2018, conducted again in August 2019 and updated in June 2020. To identify further relevant articles not included in the databases, scanning of reference lists and hand searching citations of the retrieved articles were performed. An outline of the selection process follows the PRISMA guidance which can be seen in Figure 12. The search focused on the subjective experiences of adults diagnosed with HD. Articles that explored HD lived experiences around illness diagnosis, communication, the impact of HD on everyday life and relationships were included. The Critical Appraisal Skills Programme (CASP) was used to appraise the identified literature (CASP 2014) in Appendix B.

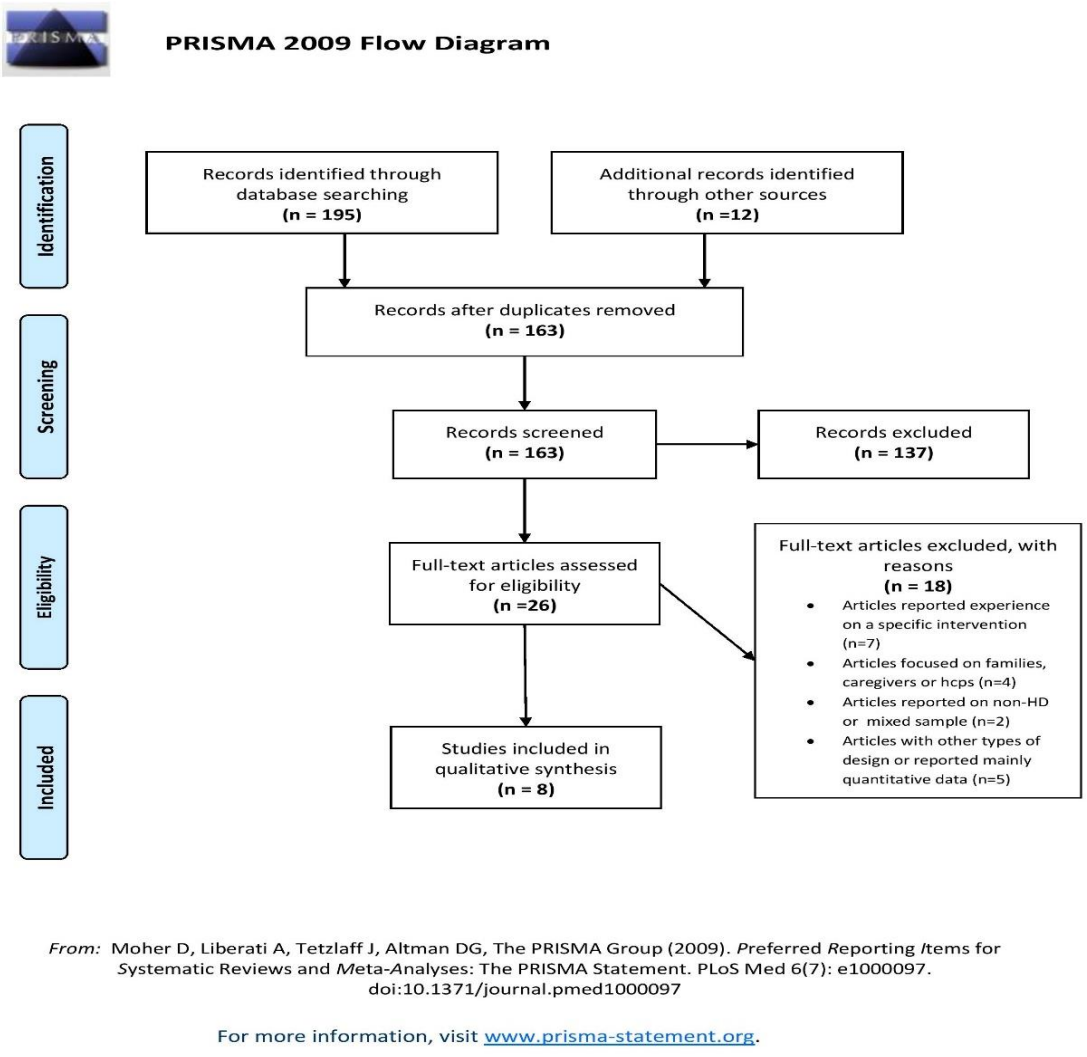


Figure 12. Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) flow diagram for the Scoping Review process

FINDINGS OF THE SCOPING REVIEW

A total of 195 records were retrieved from databases, and 12 additional records identified from other sources. The duplicates were consequently removed with 163 records screened, and accordingly, 137 records excluded. The 26 full-text articles were assessed for eligibility, and 18 excluded with reasons. A total of 8 unique articles were included in the review. See Table 3, Appendix A for the characteristics of selected studies.

METHODS AND METHODOLOGY USED

The included articles used interviews as the primary mode of data collection with one study which utilised focus group alongside the interview (Hartelius et al 2010). The dominant methodological approaches were Interpretative Phenomenological Analysis (IPA) with three studies, followed by Thematic Analysis with two studies, and Grounded theory and Narrative content analysis with one study each. Observational methods and longitudinal studies were not employed.

HD STAGES

The majority of the data collection was generated from HD patients in their early and middle stages (Hartelius et al. 2010, Ho and Hocaoglu 2011, Zarrotti et al. 2018) while the remainder of the studies did not report the participant disease stages. Further, there were differences in the description of the HD stages, and this might be attributable to the country of study and the terminology used, such that in the UK (Zarroti et al 2018) they were referred to as early, middle, and late, while in Sweden (Hartelius et al 2010), they were described as Phase 1, 2 and 3. Only one study indicated the HD stages based on the Total Functional Capacity (TFC) Scale (Ho and Hocaoglu 2011). The findings of this review revealed that research in HD late stages was rarely conducted. Quaid et al (2018) noted that apathy and cognition impairment persist in HD progression, and this may hinder active participation and the ability to engage in conversations in the later stages. A literature review in progressive neurological conditions identified that long-term planning can be difficult (Wilson et al 2008). In HD, the loss of cognition often necessitates involvement of patient caregivers in research (Wright et al 2012). Auduluv et al (2014) postulated that some sufferers from neurological illnesses were considered to be active agents (Multiple Sclerosis, Spinal Cord Injuries) while those suffering from other conditions were dependent on their caregivers, as is often the case in HD and Dementia. Wright et al (2012) suggested that people with HD appeared to have a passive role in research studies, and engaging their caregivers often generated high levels of participation and retention. The above reasons may elucidate why most qualitative studies in HD were conducted in their early stages and alongside their families and caregivers.

SAMPLING AND SETTING

All the studies above accessed HD participants through purposive sampling. This non-probability sampling method is widely used in research that is specific to the qualities of the population and their illness experience. In addition, the identification of participants' capacity and communication abilities ensured they were well informed about the research aims (Palinkas et al., 2015). Moreover, the rarity of HD has led

researchers to recruit from HD support groups and lay organisations, with one study using a social worker to recruit volunteers (Halpin, 2018). Most of the studies included here conducted data collection in participants' homes, other common settings included activity centres or the individual participant's choice of location.

KEY THEMES

The original themes and findings of the selected studies are presented in Table 4, Appendix A. This process enabled the studies' original interpretation to be maintained, and as identified by each author. This method of reporting provides important information on the selection process and offers transparency of the sampled study findings.

The selected studies revealed the HD illness experiences, and the topics explored aspects of communication (Hartelius et al., 2010; Zarotti, Simpson and Fletcher, 2019), the impact of disease on the person with HD (Ho, Hocaoglu and European Huntington's Disease Network Quality of Life Working, 2011) and family dyads (Maxted, Simpson and Weatherhead, 2014). The majority of the studies explored the genetic illness experiences; the genetic risk (Etchegary, 2011b), intersections between genes and the body (Hagen, 2018), meaning of the diagnosis (Schwartz, 2010), and dimensions "genetic suffering" (Halpin, 2018). To better understand the phenomenon of the HD illness experience, themes in each individual study were collated into a more heterogeneous group. The basic principles of textual narrative synthesis guided this approach (Lucas et al., 2007), where the sampled studies' context and findings are reported, and the commonality and the differences between studies are compared. The scoping review has applied this procedure by organising the findings using textual narrative synthesis and consequently revealed the commonality of the HD illness experience. As the synthesis progressed, these ideas were grouped together as concepts which became the basis of the major themes (Figure 13).

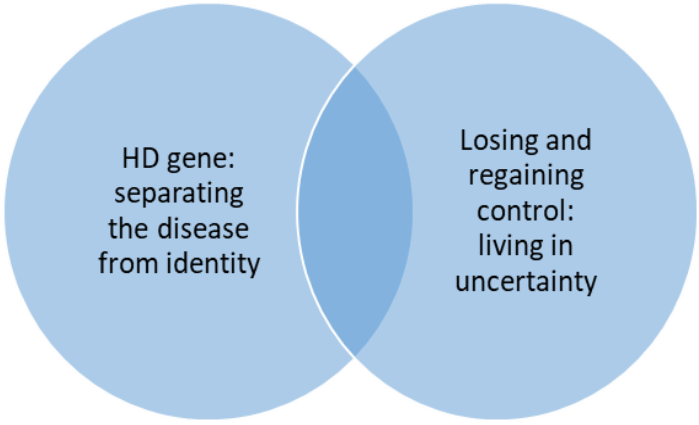


Figure 13. The overarching themes of the HD illness experience

THE HD GENE: SEPARATING THE DISEASE FROM IDENTITY

A few studies adopted a theoretical framework to underpin the individual illness experience. Hagen (2018) described the lived experience as revolving around the connections between genetics and the physical

body, as drawn from Leder's concept (1990) of the phenomenological null point (the mutated gene that causes the illness). The HD individual's everyday experience is channelled towards parts of our bodies (genes), of which we have no direct experience. Hagen (2018) observed that it is difficult for the HD individual to handle and create a stable lived experience. Hagen (2018) referenced and agreed with Etchegary's (2011) earlier study that the aspect of genetic risk is relevant to the illness experience. Etchegary (2011) proposed the chronic illness perspective (Kennan et al 2003). This helps explain the HD lived experience of the risk of illness, rather than the illness itself, a concept shared by Halpin (2018) drawing on Kleinman's (1998) distinction between 'illness', as the personal experience of the sick, and 'disease', a biological feature of the pathology. Although the selected studies provided different interpretations of how living with HD is characterised, they have in common the influence of genomics, the lay understanding of genetic risk and the experiences of genetic testing. Halpin (2018) followed theories based on science studies (Hacking 1995, Mol 2002) and medical sociological perspectives (Bury 1982, Frank 1995) and argued that genetic discourses and technologies may have a huge impact on the personal illness experience. Etchegary, (2011b), Halpin (2018) and Hagen (2018) observed that the perception of living with the illness is highly influenced by the genetic nature of HD, and that individuals tend to separate their identities from their illness. Correspondingly, when Maxted et al (2014) interviewed HD individuals together with their family members, participants referred to their relationships and identity changes as both cyclical and dynamic processes. Ho et al (2011) further emphasised that the disease impact occurs in different phases of change and stability over the protracted course of HD. Researchers further suggested that understanding the lived experience needs to be acknowledged individually (Etchegary, 2011b; Hagen, 2018; Halpin, 2018). These individual foci highlighted that there needs to be a separation of patient's identity as individuals from that of a person with a disease, which will be beneficial to maintain communicative control (Zarotti, Simpson and Fletcher, 2019) and to prevent disruptions in their social relationships (Etchegary, 2009; Etchegary, 2011b; Maxted, Simpson and Weatherhead, 2014).

LOSING AND REGAINING CONTROL: LIVING IN UNCERTAINITY

The presence of HD is perceived as something that takes over and changes the qualities of the person with HD (Etchegary 2010, Haplin 2018), communication (Hartelius et al., 2010; Zarotti, Simpson and Fletcher, 2019) and family relationships (Maxted, Simpson and Weatherhead, 2014). In people's experiences of genetic testing, the diagnosis validated the symptoms they were having as real (Schwartz, 2010), others reported a heightened level of "symptom watching" (Etchegary, 2011b). While some participants pursue the meaning of their genetic test results, for others the test results emphasised the feelings of losing control and determined the course of HD (Schwartz, 2010; Etchegary, 2011b). Some participants stated that aspects of their personality stayed the same despite their physical deterioration and loss of functional abilities (Halpin, 2018). Losing control is perceived to be one of the most substantial effects on the ability of the person with HD to communicate (Hartelius et al., 2010; Zarotti, Simpson and Fletcher, 2019). The concept of loss is a prevalent theme amongst the studies (Hartelius et al., 2010; Etchegary, 2011b; Maxted, Simpson and Weatherhead, 2014; Zarotti, Simpson and Fletcher, 2019). In addition, emotional, social and

self-identity themes were featured (Hartelius et al., 2010; Hagen, 2018; Zarotti, Simpson and Fletcher, 2019), and were present across all HD stages (Ho, Hocaoglu and European Huntington's Disease Network Quality of Life Working, 2011). Further research on how people with HD see themselves in relation to their sense of self will help contribute to a better understanding of their illness experience.

Research on HD experiences described living with genetic risk as both fluid and dynamic (Etchegary, 2011b; Hagen, 2018). People with HD described their communication as a process of struggle (Zarotti, Simpson and Fletcher, 2019) and difficult adjustment (Hartelius et al., 2010). Research on HD communication suggested that the person's inability to express emotions has negatively impacted family relationships (Hartelius et al., 2010; Zarotti, Simpson and Fletcher, 2019). Relationships in HD families either cause members to pull together or tear apart, as they constantly adapt to new roles in regards to caring and added responsibilities (Maxted, Simpson and Weatherhead, 2014). Consequently, living with HD has been described in metaphors of "under the sword of Damocles" or "spectre hanging over" (Maxted, Simpson and Weatherhead, 2014), "death sentence" or "doomsday"(Halpin, 2018). Moreover, others compare their experiences to "living with the devil" (Schwartz, 2010) or a "destructive force within" (Maxted, Simpson and Weatherhead, 2014). These illness experiences described in metaphors can help make sense of their world of living with HD and relate to the ambiguity and their feelings of fatalism (Halpin, 2018). One participant summarised this as 'whatever is going to happen will happen because it's a genetic disease' (Halpin 2018, p. 370). It is difficult and complex to create a stable personal experience of HD (Etchegary, 2011b; Hagen, 2018), as experiences are influenced by the hereditary component of the disease (Maxted, Simpson and Weatherhead, 2014; Halpin, 2018) and the impact on everyday life in different disease stages (Ho, Hocaoglu and European Huntington's Disease Network Quality of Life Working, 2011), different stages in the life course (Hagen, 2018), and disease-related events (Etchegary, 2011b).

SUMMARY OF THE SCOPING REVIEW

Most of the studies exploring the HD illness experience focus on genetic testing and diagnosis. While the genetic aspect of HD is of importance, the lack of other research topics is apparent. HD poses pervasive problems on the individual, familial, and societal level, yet very little is known of the individual experience (Halpin, 2018; Mahmood, Law and Bombard, 2021). The impact of the disease has been mainly explored by collecting quantitative data (Harding, Stewart and Knight, 2012; Mestre et al., 2018), and there remains little empirical data on people's experiences with the progressive nature of the disease.

This scoping review gave an insight into the HD illness experiences based on the available HD literature. However, the search performed did not cover all existing databases, and this consequently may have excluded papers in non-academic databases. Qualitative studies are difficult to classify using standard search strategies (Barroso et al., 2003), despite efforts to find all obtainable studies, and it is likely that some were overlooked. This can be ascribed to the inconsistent structure of how bibliographies are indexed in different search databases (Soilemezi and Linceviciute, 2018). Furthermore, not all studies provided HD patients' stage of the disease. The analysis of participants' abilities to actively communicate in

multifaceted subject areas implies they were at early stages, particularly when the topic was explored alongside their experiences of genetic testing. Only one study (Ho, Hocaoglu and European Huntington's Disease Network Quality of Life Working, 2011) acknowledged the communication needs of late-stage HD and used picture cards to facilitate discussion. Moreover, due to the wide-ranging areas of experience covered, rather than grouping the findings into distinct themes, the scoping review found heterogeneity between the studies. The main themes of the included studies overlapped, and this can be interpreted so that some findings can be interrelated. Therefore, the results need to be interpreted with caution as this does not provide an all-encompassing picture of living with HD but can only facilitate a broader understanding of the HD illness experience.

GAPS IN HD LITERATURE

The literature review has elucidated our current understanding of HD and identified the gaps in our knowledge. Moreover, the conceptual domains have structured the review into three important perspectives on HD research that have facilitated insight into the illness experience. So far, this chapter has uncovered the disease history, developments in genetic medicine and the medical management of the disease. Research on HD genetics and experiences related to genetic testing are widely studied (Quaid et al., 2008) and have dominated HD literature (Audulv, Packer and Versnel, 2014). Specifically, HD genetic testing research is unrivalled in comparison with other genetic diseases (Paulsen et al., 2013; Halpin, 2018). This aspect of the disease has established HD as a model of a hereditary brain disease, and the proliferation of research in this domain has recognised its far-reaching impact on family caregivers.

This literature review posed the same question that Aduluv et al. (2014) asked: 'Is the experience of genetic testing the most challenging aspect of living with HD or is it the most interesting aspect of HD research?' (p.204). This leads to many questions as the depth of knowledge in some areas of HD research outweighs the dearth of literature in other aspects of HD experiences. HD is a progressive disease that causes widespread suffering to individuals and families. Although our knowledge of the disease and care for individuals and families has increased over the last decade, this review identified that there is a clear gap in our knowledge of the HD illness experience. There is a significant lack of qualitative studies that seek to understand the illness experience beyond the point of receiving the diagnosis (Audulv, Packer and Versnel, 2014; Mahmood, Law and Bombard, 2021). Considering the long trajectory of the disease and the impact on the individuals' health and well-being, very little is known of peoples' experiences in the course of the disease progression.

CHAPTER SUMMARY

The findings of this literature review have contributed to the development of the main research question and study design. Moreover, these considerations have impelled the conception of the study methodology. Based on the gaps identified in this literature review, the following are identified to guide the development of the methodology and methods in this thesis to explore the HD illness experience:

1. To fill the identified gap in the literature on living with HD beyond diagnosis and genetic testing experiences and to explore the illness experience in its advanced trajectories.

2. To make use of additional methods of data collection to supplement interviews to help construct a holistic picture of the HD illness experience.

3. To explore how people with HD construct their illness experience

The study's aim is to explore the illness experience of people with HD in three manifest stages (early, middle, late) and will utilise methods of data collection tailored to the individual and their disease stage. An experiential narrative design will be guided by the research question: How do people with HD narratively construct their illness experience? The principal component of the study is the individual with HD, alongside their family caregivers and the health professionals involved in their care. By combining these multiple voices, this will help illuminate the holistic illness experience of people with HD in different disease trajectories.

Based on the findings of the literature review, this study fills a gap in HD qualitative literature and aims to focus on the voice of the person living with HD (Mahmood, Law and Bombard, 2021; Zarotti et al., 2022). It achieves this through a longitudinal approach supplemented by multi-methods and use of multiple sources of data. In addition, the study will highlight the subjective experience of people with HD in advanced stages which is rarely explored in HD qualitative studies.

The next chapter will explore the methodology of narrative research and the existing literature on illness narratives.

CHAPTER 3: METHODOLOGY: THE NARRATIVE APPROACH

In this chapter I recount my search for an epistemological position and my decision to adopt a narrative approach in studying the illness experiences of Huntington's disease (HD). Given the dearth of HD qualitative studies on the topic, I briefly discuss what we can learn from narrative studies of chronic illness and progressive long term neurological conditions (PLTNC) as this informed the study methods. Finally, this chapter concludes with a statement of the discrete aims of the study.

IN SEARCH OF A METHODOLOGY

At the study outset, I did not commit to a particular methodological framework. After the comprehensive literature review (Chapter 2), the only thing that I was certain about was that I wanted 'to hear the voice of the person with HD' as they were largely absent in the literature.

I attended several workshops and studied the five main qualitative inquiries (Creswell and Poth, 2016) to narrow my choice between phenomenology, discourse analysis and narrative inquiry. Although the work of Charmaz (1983, 1991, 2002) provided significant insights into the chronic illness experience and her approach to constructivism appealed to me, I did not intend to develop an explanatory theory of the illness experience. I considered phenomenology to understand the lived experience (Van Manen, 2016). A few researchers in our faculty were conducting phenomenological research, so I thought I could engage with them for resource and support. However, as I compared phenomenology with discourse analysis, I was also interested on the way language is used (Dew, 2007) and how knowledge can be collaborated through interaction. It is my view that Discourse Analysis loses the person and Phenomenology loses the discourse. I wanted to both capture the research interaction and maintain the person with HD at the core of the study. Through narrative research Sparkes and Partington (2003) suggest we are able 'to retain both a sense of the individual and the discourse' (p. 294). What also drew me to the narrative approach is what Frank (1995) and Reissman (2003) call the study of the particular. I wanted to better understand how each person with HD narratively constructs their illness experience.

My approach to the study is guided by narrative constructivism (Sparkes and Smith, 2008; Esin, Fathi and Squire, 2014), which has the basic tenet that reality is formed socially (Bury, 1986). As a constructionist, I believe there is no single truth or reality, rather we create multiple valid interpretations, with multiple narrative truths (Freeman, 2003). I agree with Bruner (1987) that philosophically speaking, 'narratives are means of knowing' about the world with the constructionist view of 'world making' as the principal function of the mind (p.691). Narrative knowing, according to Kramp (2014), transforms the epistemological question of 'How do we come to know the truth?' into 'How do we come to endow experience with meaning?' (p.104).

For Riessman (2003), researchers turn to narratives 'because stories reveal truth about human experience' (p.10). Stephens (2011) observed that when asked to 'describe disruptive experiences, such as illness,

people turn to stories' (p.63). Ricoeur (1991) posits that humans make sense of their lives through narratives, and Murray (2008) suggest that our sense of self is fashioned in narratives. This narrative identity is not our 'essential self' but the preferred version of ourselves in our social contexts (Riessman, 2000). Such that narratives or our life-stories do not only impart subjective information and meanings to ourselves (Ricoeur, 1991; Somers, 1994) but also reveal the broader interpersonal and social context of our lives (Riessman, 2008).

In *Search for a Method* (Sartre and Barnes, 1963), philosopher Jean Paul Sartre believed that men live with stories; 'as a teller of tales, he lives surrounded with his stories and the stories of others. He sees everything that happens through them and tries to live his life as if he were telling a story' (p. 39). Similarly, Wertz et al (2011) suggest that 'life stories link story and life, as we create stories of our lives, we live our stories' (p.65). The stories we tell others about our lives create our identity (Mishler, 2006; Bamberg, 2011; McAdams and McLean, 2013), for our stories tell who we are and about our social lives, and that culture is a 'mesh with a community of life stories' (Riessman 2008 p.10). Ricoeur (1991) describes this as the constitutive characteristic of a narrative as an intersection of the life- worlds, between the speaker and the listener, in the context of 'life as a story in its nascent state' (p.27).

For Polkinghorne (1998), people without narratives do not exist, he suggests that our narrative is 'a primary scheme by which human existence is rendered meaningful' (p.1). Equally, Bruner (1987) stated that our 'lived time' is saved in the form of a narrative, and further suggested that we recognise the meanings of our experiences through assembling them in a story form. Thus, I agree with Esin et al (2014) that a narrative constructionist approach is used by social researchers to explore 'how people story their lives' and 'understands the complexities of personal and social relations' (p.1). I would also add that people construct their reality through their stories, and that stories about our personal experience can be one of our many narratable truths (Frank, 2004; Riessman, 2008).

MY NARRATIVE TURN

After I compared different qualitative approaches (Wertz, 2011; Creswell and Poth, 2016), the study naturally aligned with the narrative inquiry as it privileges the source of storied data, highlights the individual experience, and engages in a dialogue of knowledge co-construction. While narrative research borrows some concepts from other qualitative frameworks, such as the focus on lived experience (phenomenology), or multiple data sources (ethnography), for Josselson (2011) what makes the narrative approach unique is that it 'explores the whole account rather than fragmenting into discursive units or thematic categories' (p.226) and studies the 'way humans experience the world' (Connelly and Clandinin 1990, p.2). Narratives involve practical and theoretical concepts, where the researcher is interested both in the storying process and the outcome. Polkinghorne (1998) stated that narratives must employ a meaningful structure that organises events and human actions into a 'whole' (p.18). Similarly, Reissman (1993) proposed that researchers should avoid separating stories and present them as a whole. This 'wholeness' of the narratives provides meaning and richness of experience. Bruner (1987), Polkinghorne

(1995), Reissman (1993), and Connelly and Clandinin (1990) pointed out that narratives must be organised, connected, and evaluated to translate meaning from the experience and 'as a way of honouring lived experience as a source of important knowledge and understanding' (Clandinin & Rosiek, 2007, p. 42). The storied approach appealed to me as it this can be used as a map to understand a complex phenomenon such as HD. I turned to narratives as a methodology, method and means of analysis to study the HD illness experience.

The 'narrative turn' (Atkinson, 1997; Spector-Mersel, 2010; Bruce et al., 2016), or as other researchers referred to it, the 'narrative revolution' (Clandinin, 2006; Caine, Estefan and Clandinin, 2013), 'narrative moment' (Plummer, 1995) or 'narrative wave' (Liehr and Smith, 2020), reinforced my epistemology that people live in a world shaped by stories (Bruner, 1987; Connelly and Clandinin, 1990; Atkinson, 1997; Riessman, 2000; Charon, 2012). The emerging interest in narrative research gained momentum as researchers broke away from the traditional positivist view to the postmodern research approach and the generally controlled objective boundaries of the physical sciences (Clandinin, 2006; Pinnegar and Daynes, 2007; Bold, 2011). Gubrium and Holstein (2014) argued that the narrative approach has put together the characteristics of post-structuralist, post-modernist, constructivist and ethnomethodological research. Narrative practice is the recognition of the subjective meaning in making sense of the social world, and the awareness of the researcher's reflexivity in the process.

Pinnegar and Daynes (2007) identified four changes that contributed to the narrative turn. Firstly, there is a change in the nature of the researcher-researched relationship. This change focused on interpretation and meaning, the human element of subjectivity, the relational and contextual factors and this allowed the researcher, and those being researched to learn from the study. The second was the change of research data from numbers to words. Qualitative researchers argued that by translating experiences to numeral codes, they lose the nuances of meanings, relationships, and the interest of human experiences. According to Spector-Mersel (2010) this is a result of frustration with the 'inability of quantitative methods to appreciate human experience' (p.207). The third change arises from the refocus of researchers from the general to the particular or 'the understanding of the value of a particular experience, particular setting and particular people' (p.21). Narrative researchers question the generalisation of findings and maintain that the understanding of experiences is contextual in specific places and time. The final change is when researchers turn from a single method of knowing to accepting that there are multiple ways of knowing the human experience.

These insights led me to adopt a philosophical perspective that deals with the focus on individual lives as a way to understand human existence. Individuals tell stories about their lived experiences (Ollerenshaw and Creswell, 2002) and 'these lived and told stories, are ways we create meaning in our lives, and ways we build our lives and communities' (Clandinin 2006 p. 44).

WHAT MAKES A NARRATIVE?

However, the more I immersed myself in narrative research, the more narrative became an 'ambiguous term' (Kramp 2004 p. 105). Wikan (2000) observed that 'people bleed stories, but academics gather narratives' (p.217). Stories are used as an 'informal' term in everyday discourse while narratives have 'formal' features or a certain genre (Wikan, 2000; Kramp, 2004). Moreover, 'narrative' is a term employed by qualitative researchers and draped with many meanings (Polkinghorne, 1995; Lewis and Adeney, 2014). It is a multi-faceted concept that can be difficult to define (Smith and Sparkes, 2009), as this approach does not fit neatly into specific research categories (Stanley and Temple, 2008; Lewis and Adeney, 2014), and adopts different practices depending on the methods (Riessman, 1993; Mishler, 1995) or purpose (Connelly and Clandinin, 1990; Clandinin, 2006) for individuals and social groups (Bury, 2001). Narrative may relate to the phenomenon, the process, or a research method (Connelly and Clandinin, 1990; Pinnegar and Daynes, 2007). Polkinghorne (2005) viewed narrative as 'a kind of organisational scheme expressed in a story form' (p.13). He observed that qualitative researchers use narrative as either a form of a prosaic discourse, referring to the collected data in narrative as text, or a more limited definition (Connelly and Clandinin, 1990) to a particular type of discourse, a 'story' (Polkinghorne, 2005). A story is an example of a narrative, but a narrative is not limited to a story (Kramp 2004 p. 106). Smith and Sparkes (2009) distinguished 'narratives' from 'stories' and used the term *story* when referring to concrete tales people tell, and *narratives* when conferring wide-ranging dimensions or properties, such as sequences of speech acts, structures, thematic or categorical content and/or temporality which comprise stories (p.2). However, many researchers often use these terms interchangeably (Polkinghorne, 1995; Riessman, 2000; Aranda and Street, 2001; Kramp, 2004; Gregory, 2010). Ultimately, it is through narratives that researchers try to understand the individual experience in stories (Connelly and Clandinin, 1990; Caine, Estefan and Clandinin, 2013).

In this thesis, like Frank (2010) and Kramp (2004), I will use both terms interchangeably as it 'fits the context'. Although this does not resolve the ambiguity, it does however inform our understanding of the use of the methodology (Kramp 2004 p. 106).

A BRIEF HISTORY OF THE NARRATIVE APPROACH TO STUDY ILLNESS EXPERIENCE

There has been a shift in research over the past three decades which focused on the nature of the illness experience as informed by narratives of those with an illness (Hydén, 1997; Thorne et al., 2002; Pierret, 2003; Fioretti et al., 2016; Kokanović and Flore, 2017). This emerging interest in the social sciences and medicine acknowledged the fundamental need to give voice to the patient's experience beyond the biomedical domain (Frank, 1995; Kelly and Field, 1996; Hydén, 1997; Fioretti et al., 2016). Traditionally, it is the physician's voice that holds authority over discussions around illnesses (Atkinson, 1997; Gregory, 2010)

and the speech of patients was treated with scepticism (Hydén, 1997), one that lacks the medical language to legitimately express suffering (Charmaz, 1983). Mishler (1986) contrasted these two 'voices', the voice of the people suffering and the voice of medicine and argued that often these voices interrupt one another. However, as scholars and clinicians made a distinction between illness and disease (Bury, 1982; Kleinman, 1988; Sacks, 2014), this opened a field of narrative research that recognised the illness experience as a fundamental part of understanding the disease. Notably, the physician Rita Charon (2012) advocated for producing patient narratives, arguing that conventional clinical care is not sufficient in helping individuals come to terms with their illness.

Hydén (1997) pointed out that when the focus shifted from illness to the accompanying suffering and other social contexts, this laid the foundation for reasoning that 'the patient's voice is strong enough to stand against the voice of medicine' (p.29). Since Strauss and Glaser's seminal work (1975), research interest in the meanings and experiences of chronic illness has grown, particularly in the United Kingdom (Pierret, 2003; Taylor and Bury, 2007). Strauss and Glaser's (1975) early work was instrumental in highlighting the individual's illness experience, its social impact, and the implications for public health (Taylor and Bury, 2007). Over the last three decades, this field of research has been extensively studied (Fioretti et al., 2016). The theoretical perspectives of anthropology (Kleinman, 1985; Murphy, 1998), psychology (Mishler, 1986; Polkinghorne, 1995; Bruner, 1997), and sociology (Williams, 1984; Frank, 1993; Frank, 1995; Ezzy, 1998), extend to the disciplines of medicine (Greenhalgh and Hurwitz, 1999; Charon, 2001; Shapiro, 2012), sports sciences (Sparkes, 1996; Sparkes and Partington, 2003; Smith and Sparkes, 2009) and nursing (Benner, 1991; Sandelowski, 1994; Holloway and Freshwater, 2007; Liehr and Smith, 2020), which all utilise narratives in understanding the illness experience.

The illness experience has been widely explored since the conception of Parson's (1951) sick role and the application of this concept (Gallagher, 1976; S Levine and Kozloff, 1978; Gerhardt, 1979) to the different stages of illness experience (Suchman, 1965). The 'sick role' theory introduced by Parsons (1951) is rooted in a sociological perspective of functionalism, where the delineation of health and illness and the roles of patients and physicians are derived in the context of a social system. Parsons (1951) postulated that health and illness are social experiences that are related to social roles and the expectations of individuals who fail to fulfil such roles, resulting in negative consequences for individuals and their social relationships. Despite criticisms of Parson's illness paradigm (Gallagher, 1976; Gerhardt, 1979; Kokanović and Flore, 2017), this theory has led researchers to investigate the conceptual gap between the biological, psychological, cultural, and social understandings of the illness experience.

Narrative research contrasted with Parson's (1951) concept of the 'sick person' as an object of cure which attributes the authority to the treating physician (Varul, 2010; Kokanović and Flore, 2017). Narrative approaches, specifically in a health context, have shifted this balance of power (Gregory, 2010) to focus on the individual experience (Charmaz, 1995), their personal meaning (Bury, 1982; Williams, 1984; Frank, 1993; Rimmon-Kenan, 2002) and the wider social issues (Kleinman, 1988; Kelly and Field, 1996; Hydén,

1997; Czarniawska, 2004). Sacks (1994) argued the importance of using illness narratives to inform a more detailed clinical picture of the patient. Sacks (1994), Klienman (1987) and Charon (2001) emphasise how patient narratives reconceptualise the traditional concepts of health and illness, which helped shape the contemporary biopsychosocial model of narrative medicine.

Frank (2006) articulated that health stories not only highlight the suffering of the individuals but connect them to the institutional provision of health practices. Sacks (1973) and Kleinman (1988) urged health professionals to take a more empathic approach by listening to patient stories. Likewise, Charon (2001), Greenhalgh and Hurwitz (1999) acknowledged ill peoples' narrative capabilities not only empower them, but also offer health providers diagnostic and therapeutic advantages in providing better care. Moreover, Frank (1995, 2016) emphasised the privilege of hearing a first-person account of the illness experience. Illness narratives provide personal meaning, and researchers have collected narratives of people with severe mental illness (Kleinman, 1988; Cohen, 2008), chronic heart diseases (Schwind et al., 2016), HIV and AIDS (Davies, 1997; Ezzy, 2000), spinal cord injuries (Smith and Sparkes, 2002; Sparkes and Smith, 2002; Angel, Kirkevold and Pedersen, 2009), young people who self-harm (Hill and Dallos, 2011) and older people in community care (Hsu and McCormack, 2012; Backman et al., 2018).

TYPOLOGY OF ILLNESS NARRATIVES

Kleinman (1988) has given the illness narrative concept a broader meaning in which he described how such narratives give shape and voice to the person's suffering. Similarly, Frank (1993) claimed that 'illness narratives are not illnesses but means for studying the social construction of illness 'as rhetorically and discursively bounded phenomenon' (p.41). The insights coming from Frank (1995), Kleinman (1988) and Sacks (2014) describe how narratives help people with chronic illness understand and construct their lives with modes of reasoning and representation.

In studying illness narratives, Hyden (1997) proposed three different typologies - illness as a narrative, narrative about illnesses, and narrative as illness. This suggestion was based on the relationship between the narrator, the narrative, and the illness. The first case, where illness is expressed as a narrative, is shaped by the life of the individual. This theory also relates to Labov's (1972) personal experience narratives and Kleinman's (1988) illness narratives. Kleinman (1988) stated that the personal narrative does not only reflect the illness experience but also contributes to the experiences of symptoms and suffering. The second case is narrative about illness, which conveys knowledge and ideas about the illness (Hyden 1997 p. 54). Examples include when physicians and other health care professionals talk about the patient's illness, and their narratives either guide the clinical treatment (Brody 1987), aid with communication (Clark and Mishler 1992) or support patients how to process the illness information (Good et al 1994). The final case is narrative as illness, for example, where a patient with brain injury constantly invents new narratives in the attempt to create context for his actions and his self (Sacks [1985] cited in Hyden, 1997). In this situation, there is an inability to articulate experiences in a narrative form which then becomes the basis of the suffering caused by the illness.

While Hyden (1997) classified narratives based on their formal classifications, other authors construct illness narratives based on individual beliefs (Williams, 1984) or understanding of the illness trajectory (Robinson 1990) and its relation to the individual (Frank, 1995; Frank, 1998b). Williams (1984) referred to this process as narrative reconstruction. Robinson (1990) identified the illness course as stable, progressive and regressive, and Frank (2013) classified narratives into restitution, chaos, and quest. Ezzy (2000) stipulated that different illness narratives have different theoretical emphases. In a mixed method study of people living with HIV-AIDS, Ezzy (2000) combined the different illness narratives based on the work of Davies (1997), Frank (1995), Nussabaum (1986) and Barnard (1995), and suggested that there are three different types of narratives that people use to make sense of their illness experience. The linear narratives divided into the restitutive, the chaotic, and the polyphonic. In a similar way, Kerr (2010) analysed the works of Oliver Sacks (1995) and Nancy Mairs (1996) and identified the three phases of such narratives: onset of illness, adjustment to illness and incorporation of illness into identity. Ezzy (2000) argued that while studies in health and illness privilege the linear narratives with a simple plot oriented toward stories of triumph, the multiple and often contradictory polymorphic narratives allow a person greater flexibility in adapting to an uncertain future. Moreover, Bury (2001) offered a framework for studying illness narratives, exploring three levels of narratives in the face of illness: 'contingent narratives' which concentrate on the beliefs on the onset of the disease, the proximate causes and effects of the symptoms on the body, self and others; the 'moral narratives' that provide accounts on the changing relationship between the person, the illness and social identity, and the 'core narratives' that reveal connections between the individual experience and deeper cultural meaning attached to suffering and their illness experience.

Following on from the earlier work of Gergen and Gergen (1983), Lieblich et al (1998) proposed a series of genres to underpin all forms of narrative, and more recent work by Soundy et al. (2013) elaborated these narrative types. Soundy et al (2013) highlighted these illness narratives as exemplified by different authors in the context of hope in rehabilitation for three neurological conditions: spinal cord injury, stroke, multiple sclerosis. The genres and definitions of these narratives can be seen in Table 10. According to Soundy et al (2013), the commonality of the illness narratives combines some degree of defiance, challenge, and the search for a response, for adjustment, or an acknowledgement of illness.

Table 10. Types of illness narratives (Soundy et al., 2013)

Narrative types/ genre	Definition of each narrative and examples from Author's work
Restitution	Narrative told by a patient who identifies the hope of being restored to previous state of health. Hawkins (1990) observed the desire to get well, and Frank (1995) the expectation that people prefer to hear a restitution story.

Heroic or epic	Narrative associated with social values (Kelly and Dickinson, 1997) and community ideals (Chanfrault-Duchet, 1991). Hawkins (1990) used the term 'challenge met', while Kelly (1994) described it as a courageous response to illness. Robinson (1990) distinguishes two forms of heroic narratives - the implicit: where others see it as an extraordinary way to manage illness, and the explicit: where the patient denotes 'a fight' against the illness.
Detective	Narrative that revolves around the search for answers that explore the mystery of illness. Robinson (1990) identified the need to answer the questions: Why me, and what can be done about it?
Comic	Narratives that depict illness in a different light. Patient is amused and perplexed by the illness and the response of doctors (Kelly 1994). Murray (1989) suggests conflict in a form of a comedy, and to get well through the course of medical treatment.
Ironic or satirical	Narratives that present other non-apparent outcomes. Kelly (1994) provided an example of a patient who needed surgery to save life but which had the outcome of disfigurement. Chanfraut-Duchet (1991) used the term 'Picaresque' which questions what society values, while Murray (1989) states that ironic narratives are mere schemes to control experience and believes that individuals are not so pure, and that social order is not so healthy.
Disembodied or detached	These narratives document the life events in a third-party and lack any attempt to evaluate the meaning of illness to the sufferer. The accounts whether intentional or unintentional, may lead to heroic or tragic narrative (Robinson 1990) or narratives that describe denial, such as viewing the body separated from the person (Kelly 1994).
Romantic	Narratives that are associated with a struggle for personal meaning. The narrative details a contest that a patient was involved with (Kelly and Dickinson, 1997). This narrative restores the past, highlighting a struggle to overcome a test (Murray 1989). Chanfrault- Duchet (1991) used the term 'Romanesque', which describes a quest for authentic values in a degraded world. Kelly (1994) illustrates this type of narrative with the case of a patient who stated she would get better, and that prayers from people at her church had accomplished this.
Quest	Narratives that suggest patients accept their illness and seek to use it. A sense that welcomes the future (Frank 1995) or an experience of death of

	an old self and rebirth of a new self. This is likened to the regeneration paradigm of illness (Hawkins 1990).
Supernatural or religious	Narratives that identify God as an ally, and that salvation is offered. The course towards death becomes life and losses are reconfigured into gains (Robinson 1990). Hawkins (1990) suggests three key components: life before conversion, the conversion (climax), and life after the conversion.
Didactic	Narratives that identified lessons that have been learned following the experience of illness. Kelly (1994) provided an example of disappointment in the course of the illness.
Tragedy	Narratives that illustrate an overwhelming or inevitable outcome (Bury 2001, Kelly 1994, Robinson 1990). Murray (1989) provided an example that a person can fail to conquer evil.
Sad	Narratives that represent the idea of loss and of being out of control. This narrative is not as severe or regressive as a tragedy (Robinson 1990), rather in Haskin's characterisation of a 'challenge failed' (1990 p. 551), where a patient becomes a victim of their illness.
Chaos	Narratives that are expressed in terms of loss, leaving no distance from the experience, and where the future is empty. According to Frank (1995) those who live in chaos have no reflective grasp of what has happened.

Despite its various typologies and approaches, narrative has attained a significant place in the study of the illness experience (Hydén, 1997; Pierret, 2003). Not only that narratives are used to make sense of the experience of the illness to oneself and others, but there is also evidence that this approach can have a therapeutic role for patients (Charon, 2008; Fioretti et al., 2016). Bury (1982) pointed out that illnesses cause a disruption, not only to the sense of self, but also to the individual's experience of continuity and coherence (Morse and Johnson, 1991). This discontinuation affects the fundamental aspect of life; time and its temporality (Bury, 1982; Ezzy, 2000; Harris, 2015). In addition, when a person experiences a chronic illness, this intrudes on the on-going life process as it creates a new and a different life condition (Charmaz, 2000). The individual faces the need to reconstruct their lives, to connect the past life to the present experience (Williams, 1984) and impart meaning to oneself and the world (Bruner, 1987; Polkinghorne, 1991). It is in this context that narrative becomes an opportunity to give voice to the disruption (Riessman, 1990) and a conduit for hitherto silenced voices (Hydén, 1997; Aranda and Street, 2001).

PROGRESSIVE LONG-TERM NEUROLOGICAL CONDITIONS (PLTNC) AND THE NARRATIVE APPROACH

Given the dearth of HD narrative studies, this section briefly explores how narrative approaches have been used in the cases of other neurological diseases such as PD, MS, MND and AD. These conditions share the features of progressiveness (Wilson et al., 2008), and are collectively termed as Progressive Long-Term neurological Conditions (PLTNC). This means the severity of disease symptoms increases over time resulting in a decrease in functional activity and participation. These conditions undergo 'neurodegeneration' (Schapira et al., 2014), whereby the disease pathology is primarily caused by death of neurons and other parts of the brain. HD also shares the movement disorder (Marsden, 1986) features with PD and MS, and this affects mobility and function, while the common feature with AD is brought about by cognitive and behavioural impairments. Some researchers also use the term subcortical dementias (Filley, 2019) to classify the common pathological features of these diseases, however others would dispute the usefulness of this conceptual classification (Butters et al., 1998) due to the distinctions in clinical presentations (Lundervold, Karlsen and Reinvang, 1994; Filley, 2019). Nonetheless, a substantial body of research has shown the common needs of people with PLTNC and the collective experience of psychosocial difficulties (Hartley et al., 2014; Soundy, Stubbs and Roskell, 2014), with poorer psychological functioning (DeJean et al., 2013; Arran, Craufurd and Simpson, 2014) and reduced quality of life (McCabe and O'Connor, 2012; Schapira et al., 2014; Mestre et al., 2018; Vescovelli, Sarti and Ruini, 2018).

In the selected narrative studies (Appendix B, Table 2, p.197) that were linked to the experiences of people with PLTNC, authors have used a narrative approach (as either quotes or as stories) to inform their findings. This substantiates Polkingkhorne's (1995) assertion that qualitative researchers instinctively use the term 'narrative' in their data. Some studies used the narratives pertaining to a particular inquiry, for example how narratives reveal identity construction (Hamilton, 2008b; Hydén and Örulv, 2009) or the meaning of the disease in particular contexts (Thapar, Bhardwaj and Bhardwaj, 2001; Abma et al., 2005), others presented the illness experience through general themes (Clair, 2003; Peek, 2017; Lutz et al., 2018) or storylines (Brown and Addington-Hall, 2008; Schwartz, 2010; Karlsson et al., 2014). The findings reiterate the value of eliciting narratives, most particularly to gain insight into complex illness experiences such as diagnosis (Brown and Addington-Hall, 2008; Schwartz, 2010; Peek, 2017), sense of self (Hamilton, 2008b; Hamilton, 2008a; Hydén and Örulv, 2009; Karlsson et al., 2014), communication (Clair, 2003; Abma et al., 2005), and social and physical environments (Thapar, Bhardwaj and Bhardwaj, 2001).

HOW QUALITATIVE STUDIES HELP US BETTER UNDERSTAND ILLNESS EXPERIENCE

Literature on the illness experience has proposed the theoretical analyses that provided better understanding to facilitate care and public policy (Thorne et al., 2002). Pierret (2003) stated that studying the illness experience has produced a valuable understanding of 'meanings, biographical disruptions,

narratives, sense of self and identity and coping' (p.16). The study further identified the need for social structure to be analysed (Pierret, 2003), as an individual living with chronic illness can shift their self-identity within a social context (Thorne et al., 2002). Thorne et al (2002) argued that through their extensive review of qualitative studies on illness experience, the authors did not find the 'the holy grail of chronic illness conceptualisation' (p.449), instead they encourage health researchers to orient their inquiry away from the oversimplifications of findings and acknowledge the complex and profound nature of the human phenomenon. Similarly, Audluv et al (2014) observed that more qualitative inquiry into particular neurological conditions such as HD is needed, and that research studies should employ varied methods of data collection.

Research on illness experience has engaged researchers of varied disciplines (Thorne et al., 2002; Polkinghorne, 2005). A tradition of health professionals has used patient narratives for a deeper understanding of the illness experience (Glaser and Strauss, 1964; Charmaz, 1983; Sacks, 1983; Kleinman, 1988; Charmaz, 1991; Frank, 1995; Giacomini, Cook and Group, 2000; Charon, 2001). More recently, health care services and policy makers have utilised illness stories to improve the management and delivery of health care practice and services (Pope, van Royen and Baker, 2002; Holloway, 2005; Todres, Galvin and Holloway, 2009; Braun and Clarke, 2014; Green and Thorogood, 2018). Important insights coming from the literature on illness experience were reviewed by Gerthard (1990), Thorne et al (2002), Lawton (2003), and Pierret (2003) on using qualitative evidence in the study of illness experience. Similar reviews of qualitative studies of chronic illnesses (Ambrosio et al., 2015; Joachim and Acorn, 2016) and neurological conditions (Wilson et al., 2008) support the need for empirical studies to gain deeper understanding of the illness experience. Despite the recognition of the importance of qualitative evidence in developing health policies, the proportion of qualitative literature remains very low (Hartley et al., 2014).

SUMMARY

HD literature remains dominated by clinical perspectives around the biomedical paradigm of health. While this lens was useful for HD assessment and disease management, the perspectives of family caregivers highlighted the psycho-social challenges they face. Moreover, the illness experience of people with HD has been largely absent from the literature. The scoping review in Chapter 2 identified the need to explore HD illness experiences using different methods tailored to the disease trajectory. This led to the development of the main research question: How do people with HD narratively construct their illness experience?

It is my view that the narrative approach can help us better understand the particularity and context of the HD illness experience, given that this type of inquiry has been successfully used in the cases of other chronic illnesses and PLTNC (Charmaz, 2002; Charon, 2008). In this chapter, I explained the study methodological framework and my reasons for choosing this.

This study contributes to qualitative literature on chronic disease and PLTNC experience by extending the typologies of illness narratives (Table 10 p.66) by specifically examining the experience of HD. The use of

narrative methodology on HD is currently lacking and is rarely explored in qualitative longitudinal studies. This thesis aims to contribute to the body of knowledge on illness experience (Thorne et al., 2002, Soundy et al., 2013) and the research gaps identified by Audluv et al (2014) and Zarroti et al (2022) on the need for diverse qualitative methods to explore the illness experience of people living with HD.

In the next chapter, I will provide details of the research design, the methods used in the study, the process of recruitment, and the study analytical procedure.

CHAPTER 4: DESIGN, METHODS, AND ANALYSIS

This chapter describes how the study was conducted, and it will include details of study design, the methods of data collection, the process of approvals, recruitment, and a discussion of the dialogical approach to analysis.

THE STUDY DESIGN: NARRATIVE CASES

The study adopts a case centred (Reissman 2008) design focused on the stories of three people with Huntington's Disease (HD) in different disease stages. A 'case approach' examines the individual experience and respects participants as 'subjects with both histories and intentions' (Reisman 2008 p. 12). I differentiated a 'narrative case' from a 'case study' by the unit of analysis, I focused on the participants' stories rather than a descriptive 'event, a program, or an activity' (Creswell and Poth 2016 p.78). Creswell and Poth (2016) identified several characteristics that distinguished between a 'narrative' and a 'case study', although Green and Thorogood (2018) pointed out the difficulties in classifying qualitative designs for they are 'not usually clear-cut' (p.39). Despite that, Stake (2005) and Yin (2003) offered distinctive methodological approaches of a case study as a 'methodology, method, strategy, style or approach' (Tight, 2010). Here, I have chosen to use the term 'case' as a paradigm for the exploration of in-depth narratives from a small sample of participants (Riessman, 2008; Tight, 2010).

Each case is centred on the illness experience of the person with HD (Fig. 14) and the study involved their families and health professionals. Nance's (2007) concept of the HD molecule (Fig 15) and Dahlgren and Whitehead's (1991) health determinants model (Fig 16) have influenced the study design to include in the interviews the views of family caregivers and health care professionals. Nance (2007) proposed the comprehensive approach to HD care with the patient at the centre, the outer shell composed of families and extending the bonds to multiple care providers. Dahlgreen and Whitehead (2021) offered a theoretical model to health researchers that included consideration of the various health influences and their wider determinants, a process they call 'building up the complete picture' (p. 21). The overall study design stemmed from both my experience and academic training as an HD clinician with an MSc degree in Public Health. I believe that the narrative case acknowledges the inter-connected nature of individual experience and the importance of the holistic perspective in understanding HD illness experience.

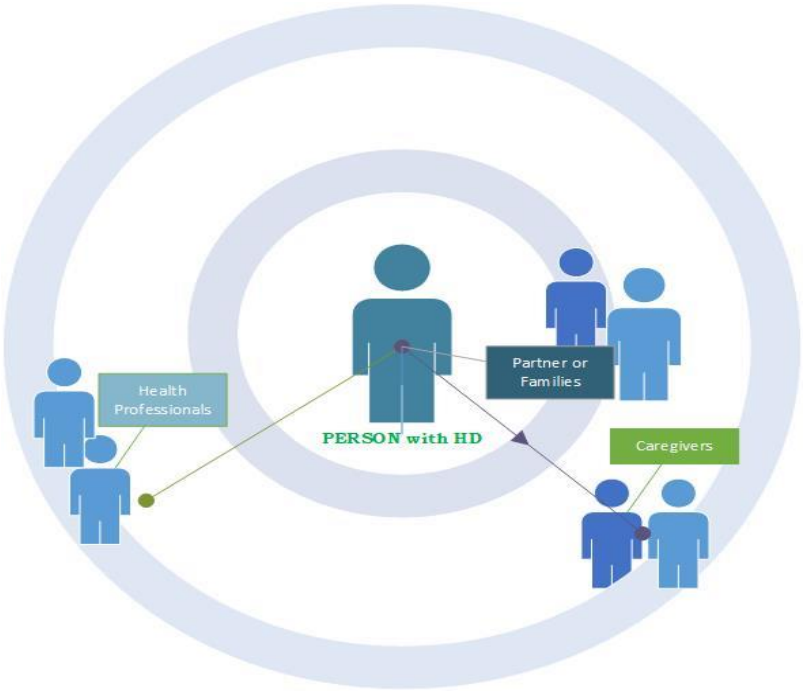


Figure 14. Narrative case design

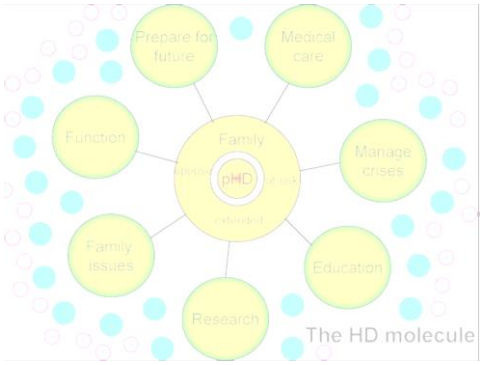


Figure 15. HD molecule (Nance, 2007)

Figure 16. Model of health determinants (Dahlgren and Whitehead, 2021)

THE PARTICIPATORY RESEARCH EXERCISE (PRE)

A PRE was held with HD stakeholders (patients, families, health professionals) before commencing the narrative cases. I felt it was important to consult stakeholders in knowledge-production (Bergold and Thomas, 2012) and adhere to a research practice that forms a collaborative endeavour, working with whoever the study is for. The inspiration for conducting the PRE stems from the disability movement agenda of *"nothing about us without us",* suggesting that stakeholders should be involved in policy, clinical care and research (Charlton, 1998; Crowther, 2007). Moreover, the PRE was my orientation to inquiry, not only did this aid my decision-making on the study methods and design, but I also gained valuable practical advice that shaped the study inquiry before I proceeded to the individual narrative cases. The objectives of the PRE were threefold, firstly this is to inform the stakeholders and generate feedback on the proposed study, secondly, I piloted the use of the interview method of eliciting a story (Wengraf and Chamberlayne, 2006) and lastly, I facilitated a discussion around perceived issues of HD research participation and gained understanding of their experiences.

The main aim of the PRE was to better understand the experiences and important issues of people living with HD, alongside the people who care for them. Literature consistently reported that little is known on the subjective experience of people with HD (Audulv, Packer and Versnel, 2014; Zarotti, Simpson and Fletcher, 2019; Zarotti et al., 2022), particularly in the advance stages of disease progression. By combining the multiple voices of people with HD, families, caregivers, and HCPs, this can enhance our understanding on the everyday life and illness experience of people living with, and affected by HD.

The PRE is of qualitative methodology design, exploratory in nature and uses focus group discussion (Bergold and Thomas, 2012) through the methods of narrative inquiry (Riessman, 2008). This has received UREC ethical approval (Appendix G). Ten participants comprised people with HD (n=2), family members (n=2), caregivers (n=3), and HCPs (n=3), were recruited through the HDA and professional networks. The participant details can be seen in Appendix C. The PRE was held in a nursing home, where the HD support group meetings were held, and the focus group lasted over 90 minutes. Data were audio-recorded and transcribed verbatim. Field notes and observations were taken. The analytical procedures were guided by two theories 1. the researcher seeking themes within the narratives (Riessman, 2008), and that experience generally involves relationships between people and contexts (Kramp, 2004).

I opened the discussion by asking the participants to introduce themselves and tell their stories about their personal experiences with HD. The focus group interview aimed to elicit a storied response, at the same time generate a naturalistic data by ensuring the participants to be more natural and relaxed in their responses(Green and Thorogood, 2018). Although there was a common initial hesitancy of where to start, when participants started talking about their experiences in a storied form, it became spontaneous, and their recollection of events automatically unfolded. Williams and Ayres (2007) discussed the importance of finding a common ground in focus groups with HD caregivers, and in the PRE this was in the form of stories. The participants started to engage and interact amongst each other. I was mindful of my role to create an

open conversation amongst participants that addressed the topic in-depth, whilst at the same time facilitating flexibility to explore often unforeseen issues that arise in the discussions. Drawing from best practices of conducting focus groups (Freeman, 2006) with neurodegenerative conditions (Jones et al., 2021) that explore multiple views of different stakeholders (Gridley, Birks and Parker, 2020), I provided each participant with equal opportunity to voice out and share their personal stories to the group. The participants naturally talked about their stories' commonalities and differences. The participants told varied stories, and their accounts were dependent on they how they were affected by HD and at what phase in their life this happened. They also spoke of their roles and relationships with the illness such that HD patients largely talked about their diagnosis period and how they currently managed their symptoms, while HD family members told stories of other family members' experiences, their caregiving responsibilities and pointed out the impact on practical resources such as time and finances. In addition, the caregivers and nurses spoke about their training and the challenges around providing care. I observed that of all the stakeholders, the family caregivers were amongst the most vocal and told emotion-laden stories about their experiences. The literature had highlighted 'the burdens' of HD family caregivers (Aubeeluck, Buchanan and Stupple, 2012) and the impact of illness on family relationships (Pickett Jr, Altmaier and Paulsen, 2007; Mantell, 2010; Domaradzki, 2015), and the PRE drew attention to their pervasive effects on family systems.

The participants were optimistic about HD research, particularly the clinical trials and gene therapies (Tabrizi et al., 2022). However, they emphasised that more knowledge and awareness are still needed for the public and amongst HCPs, particularly with GPs on the practical management and addressing the behavioural and mental health issues of HD. Family members and HCPs agree that research is necessary on how to promote a coordinated care delivery as HD interjects different health services across the illness trajectory. The two persons with HD spoke about their experiences of social stigma and their difficult dealings with health care services. The impact of HD has affected the stakeholders in different ways with no consensus on what were their most significant issues or pertinent concerns. Nonetheless, all the participants agree that one way to help improve HD care and delivery is to provide individualised care specific to the needs of the person, and that HCPs to facilitate better involvement with the person with HD in their care planning and delivery.

I gained two significant insights from eliciting stories from the PRE, this method generated detailed accounts rather than brief generalised answers. By using interviews as narrative occasions (Mishler, 1986; Riessman, 2008) participants could be encouraged to speak in natural ways and as they conveyed one story it naturally led to another. Secondly, I observed how within a group setting, the story of the person with HD gets easily drowned by other stories of family caregivers. This is because family caregivers were more articulate, vocal in expressing their emotions, and were spontaneous with their accounts. This experience taught me to be mindful in placing the illness story at the centre, as narrated and reflected by the person with HD.

ADVICE ON COMMUNICATION

The HD stakeholders gave further insight into the communication needs of people with HD, which supported the literature on the need for viable approaches to help with HD communication needs (Jona et al., 2017; Zarotti, Simpson and Fletcher, 2019; Grimstvedt et al., 2021). As HD progresses, communication difficulties arise such as problems with linguistic skills, emotion recognition and speech articulation (Zarotti, 2016). The participants suggested that HD communication issues can be addressed by personalised methods that allow a wider range of choices (Zarotti, Simpson and Fletcher, 2019) and the use of communication aids (Diehl and Riesthal, 2019; Grimstvedt et al., 2021). The nurses also highlighted the benefits of involving family caregivers as informants (Gunn, Maltby and Dale, 2020). Wright et al (2013) observed that family caregivers can help enable research data collection whilst at the same time looking after the well-being of the person with HD. HD families and caregivers agreed with this view. The two people with HD concurred, and one person with HD talked about how his caregivers helped with transport and accompanied him to participate in research in the local hospital. Moreover, caregivers and nurses suggested that when working with late stage HD patients who have lost their verbal communicative abilities, resources such as life story booklets, often used for people with dementia (Gridley, Birks and Parker, 2020) helped them get to know the history, identity and preferences of the person they care for.

I took on board the participants' views and incorporated their practical advice into my own personal experiences of communicating with people with HD. Alongside their suggestions, family caregivers and nurses emphasised that spoken words are not the only reliable means through which people with HD communicate. It has previously been observed in HD studies that despite HD communication deficits (Saldert et al., 2010; Hinzen et al., 2018), people with HD assert some level of understanding and comprehension which remains intact even during the advanced stages of the disease. The caregivers and nurses reported that the non-verbal ways of communication such as gestures and individual expressions are some of the helpful signs of communication. This was useful advice as it led me to employ alternative ways of presenting study information sheets for people in late HD stages (see Adapted Information Sheets, Appendix H).

What was particularly interesting was that when caregivers and nurses spoke about their viewpoints, they often looked for reassurance or signs of approval from the person with HD, or to the HD family members who ultimately provided some kind of approval on the validity of the illness experience. This reaffirmed my earlier observations, that ultimately the illness experience resides with the person who directly experiences it. It is paramount that the views of the person with HD remain central to this study whilst at the same time acknowledging the experiences of other people involved in their lives. This gave me impetus to adopt member checking (Birt et al., 2016) to ensure my interpretation of the data holds close resemblance to the participants' experiences.

The PRE alongside the scoping review has helped with the conception and development of the second phase of the study design (Figure 17) and this consequently strengthened the study decisions to proceed with the narrative cases to better understand the illness experience of HD. A diagram to illustrate how this contributed to the study design can be seen in Figure 17. Moreover, in contrast to conventional focus groups, the PRE approach combined with narrative inquiry provided the stakeholders a voice in the decision making on the next phase of the PhD study and the conduct of the PRE offered a high degree of reflexivity to the researcher and reflection on the research process. For Gregory (2010) narrative methodologies have a valuable contribution in participatory research as these approaches can explore participants' experience in a meaningful collaborative way and ensure participants' insights are more prominent in the research agenda.

To summarise the importance of the PRE in the wider research programme, firstly it was a useful consultation session where the stakeholders provided their views about the study and their contribution in the study design. Secondly, this highlighted the need to maintain a balanced approach in exploring the stories of individuals involved in each narrative case. Lastly, the PRE suggests that narrative methods are useful to facilitate communication and autonomy, provide choice, and a sense of control to the person with HD, to allow them to tell a personal story of the 'things that matter' (Frank 2002 p. 113).

Figure 17. Design Conception of the Narrative Cases

IMPACT OF COVID-19 ON STUDY METHODS

A couple of months after conducting the PRE, the UK government imposed restrictions (HM Government 2022) brought about by the Severe Acute Respiratory Syndrome Coronavirus 2 (Covid-19) pandemic in March 2020 (Teti, Schatz and Liebenberg, 2020). This led to changes in the research design, specifically in the case of plans to conduct non-participant observations. From March to November 2020, during the first 8 months of the pandemic, university research activities that involved in-person interventions and non-Covid related research activities were either halted or moved online. In response, the research team decided to omit the observational methods and resort to remote interviews to ensure continuity of the research project. Despite the setback, the study adapted and employed varied ways to collect data such as life story booklets (details in the preceding sections) to complement the interviews. Consequently, this opened new opportunities to explore the Voice over Internet Protocol (VoIP) and evaluate its feasibility within HD participants.

BIOGRAPHICAL NARRATIVE INTERPRETATIVE METHOD (BNIM)

Reissman (2008) observed that narrative interviews have more in common with ethnographic practice than traditional social science practices, which typically rely on open ended or fixed response questions. Mishler (1986) noted the potential problems with traditional interviews which tend to fracture the flow of conversations. He suggested that narrative interviewing could generate a sustained account in a storied form through prolonged turns at talk. I attended several narrative courses and discovered Wengraf's (2006) Biographical Narrative Interpretative Method (BNIM) as a means to invite participants to narrate their personal stories and experiences. Moreover, BNIM was particularly suited for the study design due to its longitudinal approach that asks for 'retrospective whole stories' and particular incident experiences (Wengraf, 2006; Wengraf and Chamberlayne, 2006).

The three phases of BNIM were adopted in this study (Wengraf, 2006; Wengraf and Chamberlayne, 2006). Details of the interview stages can be seen in Table 10 below. The first and second phases reflected the topics raised in the initial conversation. The participants were not interrupted, and BNIM uses encouragements such as 'nodding' and non-verbal sounds like 'hmmm' to indicate active listening. In the second stage, questions were asked in the order topics were spoken about, and mirror the language and terms used by the participants. Using this interview method provided a temporal structure that honours the stories of participants 'in their own words'. The third interview phase was used for clarification and discussion of the topics in the previous session. The use of BNIM was tailored to each participant and details of the individual interactions can be seen in Table 12, p. 86.

At the end of each interview, I also asked the participants if they were interested to read over the transcripts. In between the scheduled interviews, I either sent transcripts by post or email as an opportunity for participants to comment on their accuracy or if there were any details they wished to omit. I also asked a set of questions (Table 11) however I was aware that this might require additional time and

energy for them, or the potential to 'relive' some of their distressing stories. Therefore, I reassured them that they had no obligation to comment on or return the transcripts. Moreover, I added a fourth interview phase to discuss the 'core stories' as a co-construction process and means of member checking. I also used this time for debrief and exit interviews. I piloted this method with another PhD student I met in a narrative training session, specifically the first phase of Single Question Aimed at Inducing Narrative (SQUIN) and the second phase of Particular Incident Narratives (PINs), and it was effective in generating the flow of stories. It is my view that the use of life story interviews such as BNIM permits what Atkinson (2007) calls listening to 'one's life as a whole in the voice of the teller, as it is remembered and in a language that is deeply felt (p.237).'

Table 10. Study Interview Method with adapted BNIM (Wengraf and Chamberlayne, 2006)

Interview Phases	Study Interview Method with adapted BNIM
1	BNIM One: Single Question Aimed at Inducing Narrative (SQUIN) **'Can you please tell me your life story,** **All the events and experiences that are important to you,** **Begin wherever you like. Please take your time.** **I won't interrupt. I'll just take some notes.'**
2	↓ BNIM Two: Particular Incident Narratives (PINs) - only topics raised in session ONE - only in the order of their raising - only using the words used by the narrator *'You said… [cue-phrase] Can you remember a particular… [narrator own words] how it all happened?'*
3	↓ BNIM Three: All further questions on topics that arise in previous interviews, questions relevant to the interest and theories of the researcher
4	↓ Exit interview and obtain feedback from the core stories and participants' experiences in the research

Table 11. **Transcript questions**

Transcript questions
• Is there anything in our interview that you want to omit? Do you have any corrections to the transcripts? • Is there any information you want to remove relating to your personal information? I will remove any identifiers to ensure your privacy and confidentiality • Is there any specific information that you want to exclude or add to the transcript? I will anonymise your data, do you have any preferred pseudonym? I would appreciate any comments or feedback.

FACE-TO-FACE AND REMOTE INTERVIEWS

The decision to use remote interview methods was necessary to comply with Covid-19 social-distancing restrictions (Lobe, Morgan and Hoffman, 2020) and involved telephone and VoIP so that I could remain physically distanced from my participants. The individual details of how the study participants were interviewed can be seen in Table 4.3. For the remote interviews, the participants were provided a choice, based on their preference, accessibility to technology and how they wanted to be interviewed. Most of the participants opted for the telephone interview mainly due to its convenience, in contrast to VoIP such as the use of Microsoft Teams or Skype, which requires the use of their email address or downloading the application to their computer or mobile devices.

Fielding et al (2008) recognised the benefits of remote interviews in accessing hard to reach populations because it can overcome issues with time and space. Studies conducted during the Covid-19 pandemic by Lobe et al (2020) and Hensen et al (2020) found that remote interviews were more efficient, less expensive and less time consuming in comparison with face-to-face interviews. However, the literature around the benefits of remote interviews in the context of Covid-19 was mixed, while it was an efficient method that alleviated research practical costs (Rahman et al., 2021; Reñosa et al., 2021), others argued it restricted personal connections that can be critical in health research, and particularly in the exploration of sensitive topics (Foley, 2021; Hall, Gaved and Sargent, 2021; Hensen et al., 2021). Gruber et al (2021) suggested using multi-modal approaches and various interview modes to provide flexibility during health and social crises such as Covid-19. This study demonstrated that it is possible to adapt both remote and face-to-face interview methods tailored to the participants' needs. Moreover, the multi-modal approach overcame the barriers of pandemic restrictions which threatened the continuity of data collection.

GENOGRAMS AND ECO-MAPS

Although rarely used in research, genograms and eco-maps are used widely in clinical practice with families (Rempel, Neufeld and Kushner, 2007). In its crudest form, I used the genogram (Figure 18) to illustrate a graphic representation of HD gene lineage and family structure. Similarly, in the eco-map, I coined the term 'HD circle' (Fig. 18) to portray the social and health network of support of the person with HD. The HD circle accorded well with the narrative case design (Fig. 19) as the diagram gave a visual image of the people involved in the social and health aspect of the HD individual.

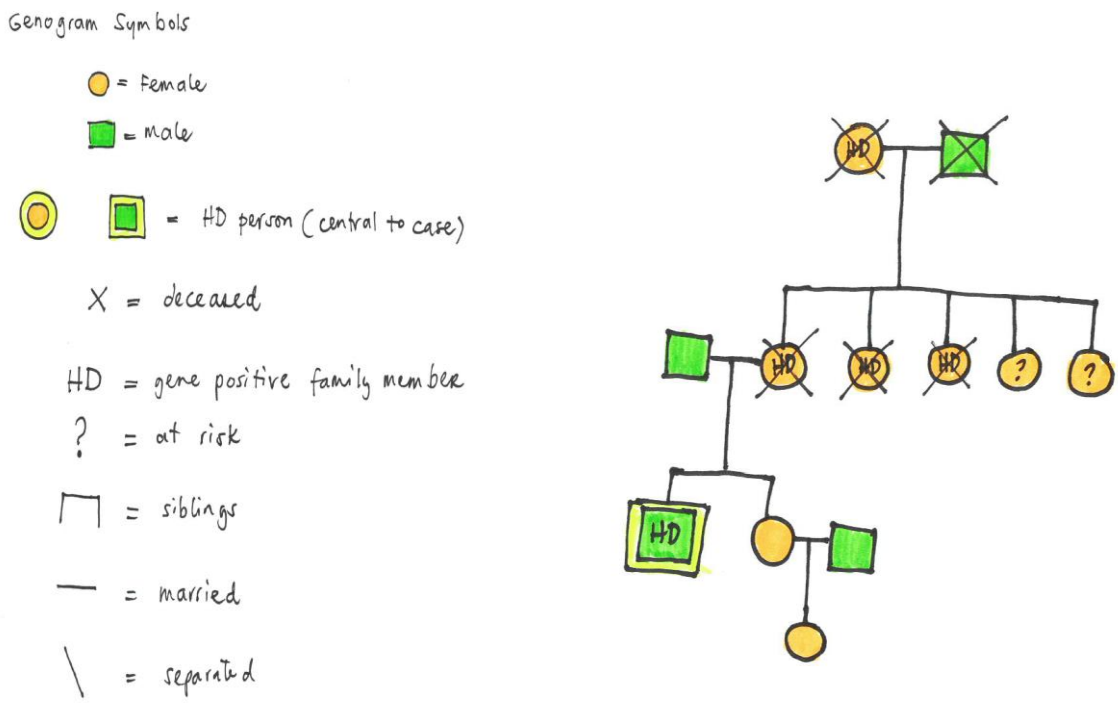


Figure 18. Genogram of a Narrative Case

A few HD studies have utilised genograms to better understand the relationships between grandparents and grandchildren (Armas Junco and Fernández Hawrylak, 2021) or situate implications of DNA testing within family systems (Brouwer-DudokdeWit et al., 2002). In a study on family caregivers, Rempel et al (2007) used both genograms and eco-maps to understand each family member's experiences. Manja et al (2021) advised that the use of eco-maps can be adapted to meet health research needs, and when used in conjunction with interviews they can promote data credibility through triangulation. Likewise, Olsen et al (2004) state that by blending the central concepts and use of pedigrees, genograms and eco-maps can help communicate a holistic picture. In this study, I used genograms and the HD circle to complement the interviews, to envisage how the HD gene runs through the participant's family and locate the relationships and sources of support of the person with HD. These tools were hand drawn by myself and I presented them alongside the interview transcripts for participants to verify and comment on.

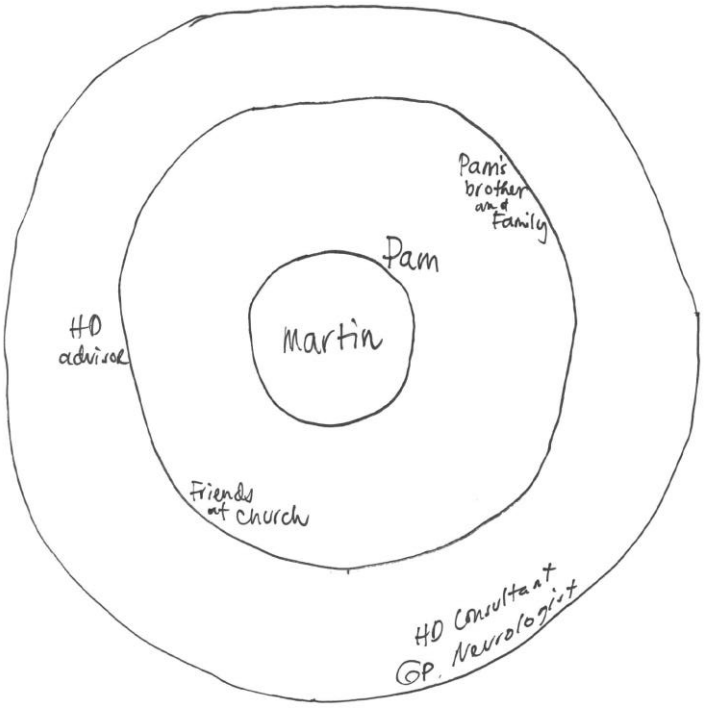


Figure 19. HD Circle exemplifying the HD social and medical network of support

LIFE STORY BOOKLETS

I devised a life story booklet as an additional research tool to generate further data that also complies with the Covid-19 restrictions. I was trained in and familiar with the use of this resource when I worked as an Activities Coordinator in various EMI (Elderly Mentally Infirm) care homes. The life story booklets were also suggested by HD stakeholders in the PRE as a useful means to get to know the person with HD. I condensed and adapted the freely available online resources from Dementia UK (https://www.dementiauk.org/wp-content/uploads/2017/04/Dementia_UK_Life_Story_Template-April-2017.doc) and Age UK (https://www.ageuk.org.uk/bp-assets/globalassets/oldham/new-content/documents/life-story-form.pdf). The version I used in the study can be seen in Appendix D.

At the time of the pandemic, I sought an appropriate method to continue with data collection while mindful that the remote methods should also decrease the emotional risk and distress in recalling past experiences. Although not specific to HD nor to Covid-19, dementia studies have reported positive outcomes on the use of life story materials (Subramaniam, Woods and Whitaker, 2014; Elfrink et al., 2018), including improvements in mood and cognition (Morgan and Woods, 2010) and developing a sense of connectedness (Kindell et al., 2014). I remembered working with people reminiscing and writing their life stories in a guided template, and that they enjoyed the activity as it gave them a sense of control over the process of what to write. Kindel et al (2014) observed that when stories are 'recalled and reframed', the past (or present) 'distressing life events become more palpable to enable everyday functioning' (p.151). My personal experiences with life story work gave me confidence to send the documents through the post

and consequently advise the participants to fill it in their own time, by themselves or with the assistance of their caregivers.

OTHER DOCUMENTS AND ARTEFACTS

Denzin (1989) recommended using personal documents for studies that include a biographical perspective. However, he cautioned that using personal documents can only represent partial identities and lives and should be analysed and treated as such. Bloomberg and Volpe (2018) state that narrative inquiry relies on different methods such as interviews, text data, fieldnotes and other artefacts such as 'photos, videos, artwork, journals, or letters' (p. 97). Hence, I utilised these available narrative sources to generate and complement each other for a holistic view.

However, I did not seek specific documents or artefacts, rather I relied on what participants wanted to share or mentioned in the interviews. The artefacts varied between participants, examples included paintings, family photographs, lists of medications and hospital letters. Congruent with the narrative methodology, the methods used in the study encouraged the participants to tell their story, in their own terms, in their own ways (Riessman, 2008; Frank, 2012).

FIELD NOTES AND RESEARCHER JOURNAL

I recorded in my field notes details of each interaction and wrote guides to inform the direction of the next interview. The entries varied considerably, ranging from sketches of interview settings to noting the words that participants put emphasis on.

Before I embarked on each research interview, I reflected and ensured that two main questions were answered: 1) Will their participation in the interview cause undue stress? 2) How can I facilitate a safe space for participants to talk? While the risk assessments mitigated the physical risks of Covid-19 (Appendix M), I had to resort to the literature, and my own lived and professional experiences in handling sensitive issues and emotional risks posed by the study with the additional impact of Covid-19. After each interview, I wrote in my research journal any reflections as a debrief and act of self-care to detach myself from the stories of the participants. My thoughts would focus on 1) my feelings about the conduct of the interview 2) a lay summary of the content of our discussions 3) observations on participant non-verbal signs and which part of their story had they put emphasis on 4) my reactions during the telling of their stories 5) the areas of discussion that needed further exploration 6) anything I felt I should have or could have done better and lastly, 7) any action for me to resolve or needed to act on as a result of their disclosure. Moreover, I would immediately debrief with my supervisors either through face-to-face, video call or email.

These notes not only helped me in visualising and remembering the details during the data analysis, but also helped me with the process of self-reflexivity, as a means to understand my role and thoughts as a researcher (Sword, 1999). Example entries can be found in Appendix E.

TOO MANY DOCUMENTS, TOO MANY METHODS

Kim (2015) referred to a 'cabinet of curiosities that represents wonder' and posits that collecting different documents and artefacts is an 'important narrative method that narrative inquiries should engage' (p.178). Reissman (2008) provided a useful discussion on how exemplary narrative studies used different documents and how they interpreted images alongside spoken and written texts. Reissman (2008) and Kim (2016) contend that the diversity of narrative methods requires the researcher's creativity and imagination to find meaningful data. Benson (2014) noted that when researchers used multiple sources of data, it is 'difficult to do justice to all data collected' (p.158), however he recognised the advantage of triangulation in that one source can be tested against another. Flick (2018) provided a useful definition of triangulation and its contribution to research quality; he advocated the use of multiple data and multiple sources as means to enhance the study findings.

In this study, using different methods made it easier for participants to share their stories and allowed me to cross check the details. In addition, I provided each narrative case its own 'physical box' akin to Kim's (2015) version of 'cabinet of curiosities'. I also created an electronic version for back-up with scanned copies. All field notes, observations, and other notes, such as email correspondence, phone calls and my observations about the participants were included. I included in the narrative case box anything related to the case interactions, including my own thoughts and interview notes. This system not only functioned as storage, but also helped me in the analysis stage where I could visually arrange data to help create the story of participants' illness experiences.

PROCESS OF APPROVALS

The study underwent several University Research Ethics Committee (UREC) applications centred on the issues around study methods and design. According to UREC, the study was deemed bearing a high level of risk due to the sensitive nature of the research. However, the study did not require NHS REC as the research and recruitment of participants did not take place in NHS settings or affect any NHS service. The protocol was checked against Health Research Authority and NHS Research Ethics Committees (RECs) guidelines https://www.hra.nhs.uk/approvals-amendments/what-approvals-do-i-need/hra-approval/. The flow chart of the process of approvals can be seen in Appendix G. The initial applications were not granted UREC approvals as the committee required further evidence to support the safety of the participants. The amendments included more information on consent process, limits of confidentiality, and risk assessments to ensure participants were not harmed in the process. The summary of the approval process, details of the amendments and provisos can be seen in Figure 20. The first ethical application was deferred, with the UREC recommending the provision of separate applications of the PRE and case studies. Approval for the PRE was applied for in April 2019 and granted in June 2019. The second ethical approval was granted in December 2019 to commence with the narrative case studies. The final ethical application was made in

June 2021 to the resume with face-to-face data collection at the time of Covid-19 pandemic with subsequent risk assessments and safety measures (Appendix M).

The process of obtaining approval was lengthy and complex. Each phase of the study presented its own set of ethical issues as seen in Figure 20. Each application was amended, the majority of the UREC provisos were centred around procedural issues and risk management. The UREC provided specific conditions and advised that any adverse events or unforeseen ethical issues should be immediately reported to the committee.

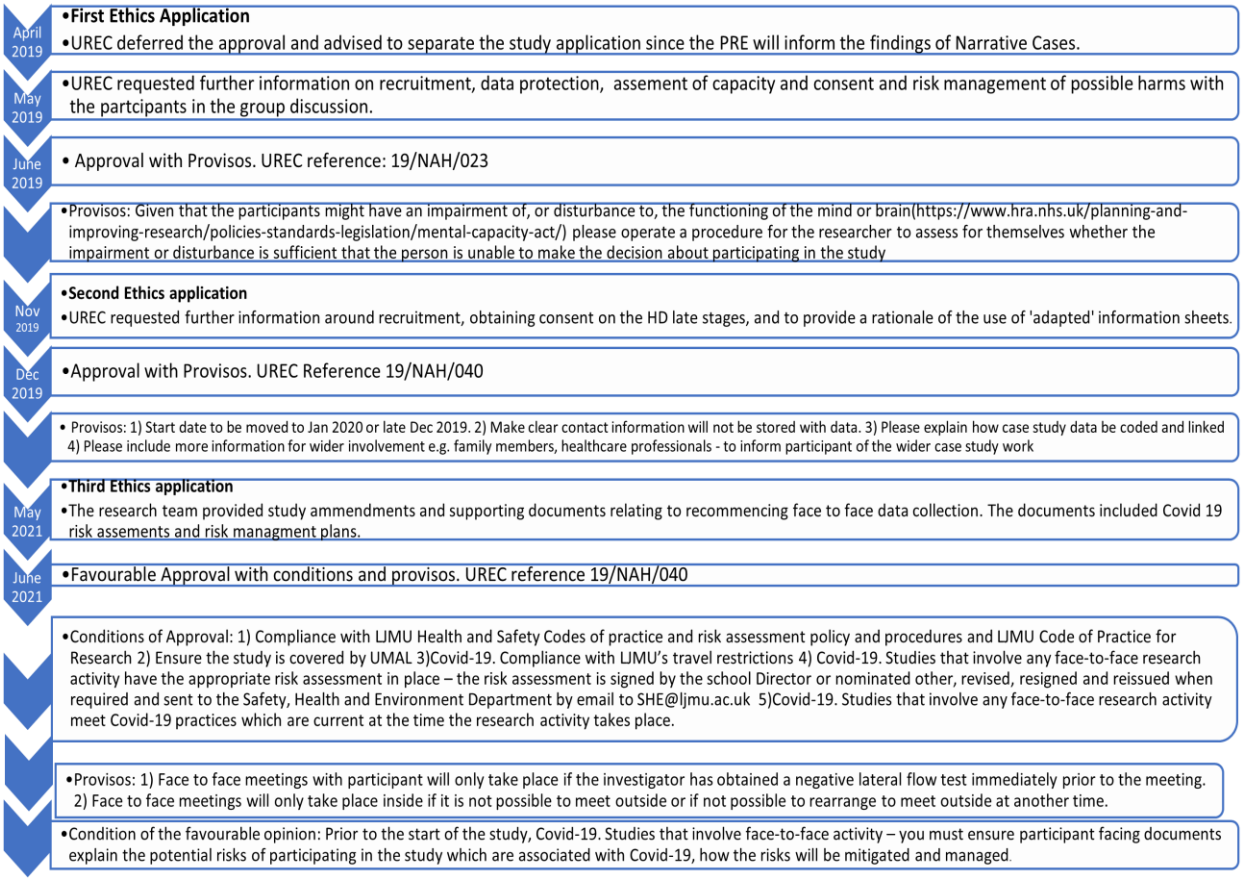


Figure 20. Summary of Ethics Application and Approval Process

ETHICAL CONSIDERATIONS

CONSENT

Obtaining consent is an important issue in health research and has been the cornerstone of most ethical guidelines (Green and Thorogood, 2018). This is a particular challenge in HD, given the disease symptoms can manifest fluctuating capacity relating to issues around physical and/or cognitive domains. Hence, I attended to these issues by adapting information sheets and consent forms (Appendix H) based on Wilson et al's (2010) pragmatic approach and use of reader-friendly information sheets. They were specifically designed to address the HD physical and cognitive effects. The HD forms were drawn from relevant

literature that adheres to the principles of Mental Capacity Act (MCA) 2005 and that encourages people's decision making (McKeown et al., 2010; Parker, Penhale and Stanley, 2010). Wilson et al (2010) suggested the inclusion of key elements to aid with understanding such as the use of large fonts, pictures, and accessible language with lots of white spacing. I also draw elements from the literature on people with learning and communication difficulties and their participation in narrative studies (Atkinson, 2004; Cameron and Murphy, 2007; Hamilton et al., 2017) to aid with the study information forms. Despite the lack of research on the feasibility of these information sheets, this study has proved the effectiveness of the adapted forms with HD participants.

After obtaining written consent, further verbal consent was sought at the beginning of each research interaction. As I viewed consent as a continuous process rather as an irrevocable decision, I reminded participants periodically that they could withdraw from the study at any time, without giving me a reason. The consent forms and information sheets can be found in Appendix H. They can also be accessed via the study website https://voicesofhuntingtons.wordpress.com/participate/.

CAPACITY ASSESSMENT

Details of capacity procedures in health research are rarely reported (Jimoh et al., 2021). One of the key issues highlighted by the UREC is for the researcher to operate a procedure that assesses the participant's decisional capacity to participate in the study. In addition to information sheets, I corresponded with gatekeepers to ensure potential participants had been assessed to have capacity and that they met the study inclusion criteria. Moreover, I implemented a protocol (Appendix J) in line with the Mental Capacity Act (MCA) (HM Government 2005) incorporating my experience as a mental health clinician. In addition to the assessment of gatekeepers and HCPs, I assessed the capacity of the person in relation to a specific decision, in this case their ongoing research participation. The HD participants, regardless of their stages, were able to demonstrate capacity in understanding and retaining information and communicated decisions relevant to the study and their participation.

Researchers have argued that while the MCA safeguards people with capacity issues from intrusive research, the Act does not account for the diversity of social research and often hampers studies from taking place (Parker, Penhale and Stanley, 2010). Obtaining consent and capacity which stemmed from the model of biomedical research is often impractical and inappropriate into social research, for it has little if any risk of physical harm (Wilson, Pollock and Aubeeluck, 2010). Moreover, a systematic review of studies relating to MCA and participant recruitment found that people with cognitive and communication difficulties are often excluded on the basis of capacity and communication issues, and that they continue to be under-represented in research (Jimoh et al., 2021). To overcome these barriers in an already under-researched population, what helped the HD participants with recruitment was the comprehensive methods of communication with the participants, liaising with gatekeepers, the use of adaptations (Jimoh et al., 2021), and the development of new resources to support consent and capacity decisions (Cameron and Murphy, 2007; Wilson, Pollock and Aubeeluck, 2010; Hamilton et al., 2017).

CONFIDENTIALITY

Ethics in social research underline confidentiality as a key criterion for ethical practice (Green and Thorogood, 2018; Association, 2021). One of the challenges was that the HD families involved lived in a narrow geographical area and worked with a small network of HCPs, leading to possible identification of individuals through the stories they told. Participants were informed of these issues and the limits of confidentiality were discussed in the study information and consent forms. I tried to manage and minimise these issues of potential indirect identification by working with the participants, families, and HCPs through removing personal identifiers, use of pseudonyms and by having discussions on what details to exclude in the narrative findings. Confidentiality was achieved by changing any personal data without altering the essence of their experience or affecting the relevance or impact of their narratives.

Moreover, participants were invited to read the interview transcripts and review the core stories (McCormack, 2004) before feeding back on any details they wished to omit or change (Table 4.2). In this way I was able to write thick descriptions (Ponterotto, 2006) without compromising anyone's identity.

RECRUITMENT

The study was granted ethical approval for different recruitment streams. Recruitment was made possible through links with HCPs and the HDA Charity. Due to disease rarity, I decided to first approach the HDA charity for advice. I contacted the HDA Service Manager through email and informed her about the study and recruitment plans. The manager accordingly requested copies of ethics approvals in order to endorse the study and I was advised, since I intended to meet participants in person, that the best way to recruit was through the local HD support group and to contact the regional HD advisor who could act as a gatekeeper.

The aim was to explore the illness experience centred on three people with HD at different stages. I was given several opportunities to talk about the study in the local HD support group, and I was able to recruit one HD participant this way. Respectively, the other two HD participants were recruited through the HD advisor and through my previous workplace. Details of how I met each participant are elaborated in Chapter 5.

The study recruitment flow process can be seen in Fig. 21 below. Although it could be argued that this process posed an element of bias, as the participants were connected with HD organisations or identified by HCPs, I deemed it important to find suitable participants and for the study to remain close to HD networks. These ensured the study risks were mitigated given the longer study duration and the emotional risks involved in discussing sensitive topics. Moreover, by keeping in close contact with HD support groups, this can provide access and timely support, should the participants need them.

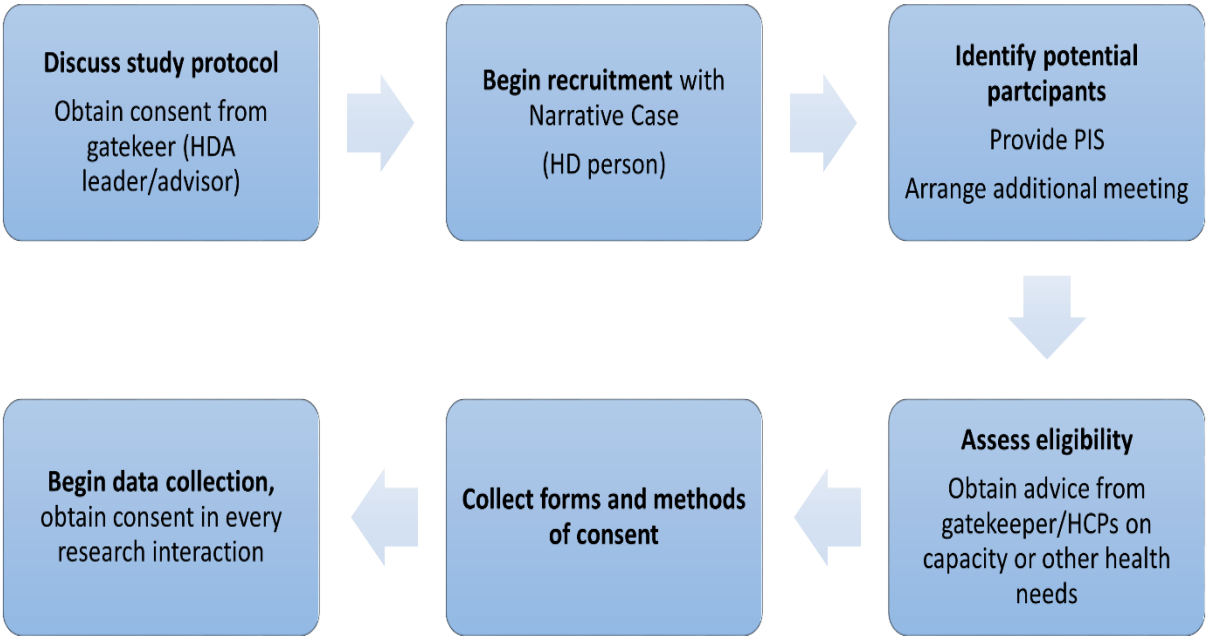


Figure 21. Study Recruitment Process

Once the person with HD met the study criteria (Appendix I) and was determined by the gatekeeper and/or HCPs as eligible to participate in the longitudinal study, they were given study information sheets. I advised potential participants to ask questions and to take time in reading the study details. Moreover, if the person with HD wished to involve their families or HCPs in the study, they were provided with specific information sheets (Appendix I). In the period of Covid-19 restrictions, participants were given the option to download forms from the study website, or to have them sent by email or by post. In addition, electronic signatures were accepted, and email correspondence was recognised as proof of consent.

RECRUITMENT ISSUES

Issues with recruitment were observed during the period of UK Covid-19 restrictions, such as losing contacts with participants who initially expressed their interests. One participant dropped out of the study, another participant had experienced recent psychiatric issues and another two participants who had significant health issues informed me that they could no longer commit to the study. This echoed the issues of recruitment and data collection experienced by other doctoral researchers during Covid-19 pandemic (Lambrechts and Smith, 2020; Campbell, 2021), highlighting the challenges around participants' health issues (Cardel et al., 2020), time constraints and social distancing (Tremblay et al., 2021).

STUDY SAMPLE

Three people with HD participated for the entire study period of 18 months. Each narrative case corresponded to the classification of the early, middle, and late HD stage based on the person with HD Total Functional Capacity Score (Chapter 2, Table 4). The HD participants decided to either involve their families or HCPs. The visual representation of the narrative case, people involved in the case and the methods used in study can be seen below (Table 12). In total, there were nine people involved in the narrative cases.

Narrative Case one is centred on Jen Walters with late HD stage currently living in a nursing home. Her case involved her children, Timothy and Marjorie Walters, her social worker Poppy Williams, and the accounts of her nurse Maria Crowley. She received her HD diagnosis over ten years ago.

Narrative Case two is focused on Martin Taylor with middle stage HD. Also involved in the study is his wife, Pamela Taylor. Martin received his diagnosis three years ago. The couple reside in their family home. They have no children.

Narrative Case three concerns Daniel Carragher, a single man with early HD stage. He was referred to the study by the HD advisor, Amanda Watts. Daniel received his diagnosis over twenty years ago. He was the main caregiver to his mother, from whom he inherited the HD gene. His mother passed away at the beginning of the study. He currently provides care for his elderly father in their family home.

Table 12. Summary of the narrative cases, the people involved in the study and methods used

Narrative Case (NC)	**NC1. Jen Walters**	**NC2. Martin Taylor**	**NC3. Daniel Carragher**
HD Stage	Late	Middle	Early
TFC Score at start of study	2	7	12
TFC Score at end of study	1	5	12
Age of HD diagnosis	43	65	25
Age at start of study	56	67	44
People involved	Children (Timothy and Marjorie) Social Worker (Poppy Williams) Nursing home staff (Maria Crowley)	Wife (Pamela Taylor)	HD Advisor (Amanda Watts)

Interviews/date **F (Face-to-face)** **T (Telephone)** **V (VoIP)** **Joint Interviews (J)**	21.02.20 (F) 21.09.20 (F) 06.01.21 (T) 17.02.21 (T) 11.05.21 (V) Timothy: 23.06.20, 05.06.21 (T, V) Marjorie: 07.07.20 (T) Poppy: 16.07.20 (T, V) Maria: 21.02.20 (F) 13.10.21 (F)	29.01.20 (F) (J) 19.02.20 (F) (J) 09.07.20 (T) (J) 13.05.21 (T) (J)	26.05.20 (T) 13.07.20 (T) 15.12.20 (F) 13.05.21 (T) Amanda: 27.05.20 (V)
Exit interviews	13.10.21 (F)	11.10.21 (F)	12.10.21 (F)
Genogram and HD Circle	✓	✓	✓
Documents and Artefacts	✓	✓	✓
Life Story booklet	✓	✓	✓

STRENGTHS AND LIMITATIONS OF STUDY METHODS

The small sample is one of the defining features of this narrative study (Creswell and Poth, 2016) for it concerns itself with the quality of the sample and the depth of the analysis (Riessman, 2008, Josselson, 2011). Moreover, the multi-methods used in this study are not suitable for large numbers, for example the BNIM method requires extensive interviews over a prolonged period of time (Wengraf, 2006). In this study, the careful selection of each HD narrative case is worth documenting and analysing due to the rarity and complexity of the illness. The extensive participant data captured the changes in experiences and perspectives over time. The study methods and longitudinal approach were essential to fully understand the unique illness experience of the disease progression and the impact of their health interventions.

Moreover, the longitudinal approach provided rich, detailed data that can be used to develop a nuanced understanding of how the disease affects individuals and families (Andersson et al., 2016) and was critical to answer the study question of how people with HD construct their illness narratives.. In addition, the longitudinal study allowed room for flexibility (Henderson et al., 2012; Whiffin et al., 2014) as I was able to modify the methods of data collection during the pandemic and learned more about the participants, This

is particularly important in studying a disease such as HD, for it can have unpredictable effects on individuals (Craufurd and Snowden, 2002; Ghosh and Tabrizi, 2018).

There were also several limitations in conducting a longitudinal qualitative study for people with HD, which included intensive demands on time, lack of resources and challenging recruitment of participants which made the study difficult to conduct, particularly in the context of rare diseases (LaDonna and Ravenek, 2014). Moreover, longitudinal studies raise ethical considerations related to participant confidentiality, privacy, and autonomy (Etherington, 2007; Wilson, Pollock and Aubeeluck, 2010; Andersson et al., 2016). Therefore, I applied high levels of sensitivity to the potential vulnerabilities of individuals with HD and their families and took appropriate steps to ensure that their rights and dignity are protected throughout the study.

FINDING THE APPROACH TO ANALYSIS

I tried to look for a specific narrative approach to analysis that was in harmony with the research question, and which was congruent with my epistemology. Reissman (2003) referred to narrative research as a 'family of methods' that have a common storied form, but as in all families, there can be conflicts, disagreements, and different perspectives (p.11).

Mishler (1995) and Reissman (2002, 2008) elucidate how narratives can be used differently in the analysis of structural forms, themes, dialogical and performative functions. A number of scholars have presented different models of analysis (Connelly and Clandinin, 1990; Mishler, 1995; Lieblich, Tuval-Mashiach and Zilber, 1998; Riessman, 2000; Sparkes, 2005; Cortazzi, 2014), while some focus their analysis on structure and form (Robinson, 1990; Riessman, 1993; Frank, 1995; Labov and Waletzky, 1997), others on content (Davies, 1997; Ezzy, 1998; Sparkes and Smith, 2003), performative (Riessman, 1993) or dialogical (Riessman 2008, Frank 2010) aspects of the story (Crossley, 2003; Riessman, 2003; Holloway and Freshwater, 2007).

While Lieblich et al (1998) and Smith and Sparkes (2009) each offered a framework for analysing the broad principles of narratives, it is also worth pointing out that narrative analysis is not guided by sets of formal rules (Riessman, 1993) nor does it attempt to sanitise research by following scientific process and linear trajectories (Fraser, 2004). As Coffey and Atkinson (1996) stated 'there is no formulae or recipes for the best way' to analyse narratives (p.80). Bleakely (2005) likened narrative analysis to 'more art than research' and suggested that it 'can hardly be taught' (p.537).

As Sparkes (2005) pointed out, one of the strengths of a narrative approach is that it 'opens up the possibilities for a variety of analytic strategies' (p.194). Likewise, Smith and Sparkes (2009) stated the need for analytical diversity in narrative research. Indeed, researchers have illustrated diverse ways of analysing narrative data (Mishler, 1986; Riessman, 1993; Mishler, 1995; Polkinghorne, 1995; Labov and Waletzky, 1997; Lieblich, Tuval-Mashiach and Zilber, 1998; Riessman, 2000; Ollerenshaw and Creswell, 2002; Fraser, 2004; Sparkes, 2005; Crossley, 2007; Harbison, 2007; Frank, 2012; Cortazzi, 2014; Esin, Fathi and Squire,

2014). Further examples include studies by Frost (2009) which combined several analytical techniques and used the term 'within-method pluralistic approach' (p.9), whilst McCormack (2004) utilised multiple lenses of visual and textual strategies to construct the participants' stories. The combination of different narrative analytical techniques has made an important contribution to the understanding (Smith and Sparkes, 2009) of the varied meanings of an individual's world (Frost, 2009). Reissman (2008) emphasised that whatever mode of analysis the research undertakes, it must be in line with their research aims, their understanding of the phenomenon, and disciplines of the study.

TAKING A DIALOGICAL APPROACH: ARTHUR FRANK'S STORYLINES

Sparkes (2005) summarised narrative approaches in the context of health research and Arthur Frank's (1995) work has been highly influential in the study of illness narratives. Frank's theory was largely centred around the illness storylines of Quest, Chaos and Restitution (Frank, 1995) and the dialogical aspect of narratives (Frank 2010). Frank's illness typology has been a focal point for numerous studies ranging from work on the experiences of living with HIV (Ezzy, 2000) with stroke (France et al., 2013), and more recently with Covid-19 (Rushforth et al., 2021).

My critique of literature that referenced Frank's typology can be seen in Table 4, Appendix B, p.204. Certainly, other scholars have critiqued Frank's approach, for it lacks clarity and rigorous analytical process, and relying heavily on the researcher's interpretation (Atkinson, 1997; Strawson, 2004; Woods, 2011). One can get caught in between methodological debates in the contested terrain (Thomas, 2010) of how illness narratives should be studied (Frank, 2000; Bochner, 2001; Mishler, 2005; Atkinson, 2009). Frank's book, *The Wounded Storyteller* (Frank, 1995), changed the way I viewed illness stories reminding me that I need to 'hear stories to know, as they are known' (Frank 1998 p.199). I want to participate in a research endeavour that Frank (2002) suggests 'goes far beyond production of knowledge from and about people's stories, and to use stories 'toward something better' (p.116). Frank's next work *Letting stories breathe* (Frank, 2010) put emphasis on the dialogical shift in research (Frank, 2012) that voices tell stories and stories can be vital sources of empirical knowledge (Frank, 2010).

In contrast to thematic approaches, Frank (2002) argued that 'narrative analysis begins with an attitude towards stories' (p.113) and 'less about finding themes and more about asking what stories do' (p.2). Frank's archetypal storylines of Quest, Chaos and Restitution (Frank, 1995) and Dialogical Narrative Analysis (Frank, 2010, 2012) was seminal to this study as it grounded a theoretical reference point for understanding and analysing HD illness narratives. Frank's approach focuses on context, and it reflects narrative elements of the story content and form. Frank (2009) and Reissman (2002) maintain that the study of narratives begins with putting a human face to a story for people who need to hear them. I took this approach to better understand the HD illness experience through stories as described by participants in their own voices. Hence, my study was founded by this 'respect' for stories (Frank 2002 p. 115) and their capacities to construct and explain, to help readers better understand the illness experience (Frank, 1995).

In *The Wounded Storyteller*, Frank (1995) proposed three storylines found in illness narratives: Quest, Chaos and Restitution. He contends that the boundaries between these three forms are not always distinct, that they are nested at different illness stages and that most stories combine elements of all three. For Frank, Quest (the search for answers and a sense of purpose), Chaos (a sense of threat, disorder, and disruption) and Restitution (the restoration of former sense of self) underline the plots and tensions of illness narratives. Frank (1995) cautioned that storylines do not give a unifying view of the illness experience, rather they can be used as 'listening and storytelling devices' to weave different narrative threads and for readers to appreciate the 'particularity of the individual experience' (p.76).

QUEST

The Quest storyline provides the person with a distinct voice to their illness experience (Frank, 2012). Frank (1995) observed that most published illness narratives are about a quest: illness is lived as quest to gain new insight (Frank 1998, 2002). What is quested for may 'never be wholly clear,' but the quest is defined by the ill person's belief that 'something is to be gained through the experience' (Frank 1995, p.115). There are three facets of quest narratives: (1) memoir, in which events are recollected and incorporates illness into one's life; (2) manifesto, where the story demands social action or change; and (3) automythology, which describes the process of self-reinvention that reveals an individual change (Frank, 1995). Similar to the concept of 'quest stories leading new insights' (Frank 1998 p.203), Williams (1984) drew a conceptual strategy of a 'narrative re-construction' (p.175) from his thirty case studies on people with rheumatoid arthritis. Narrative re-construction describes the means by which people change their relationship with the world, caused by their 'biographically disruptive' illness (Williams, 1984 p. 177). They 'reconstitute and repair ruptures between body, self and world' and they quest to 'realign present and past' (p.197). For Ezzy (2000) quest narratives are the ill person's heroic attempt to orient to the future. Frank (1995) suggests that illness as the occasion of a journey becomes as quest. The characterisation of illness as a journey is a familiar metaphor in studies on chronic illness (Little et al., 1999) and is associated with a heroic narrative positively constructed where the individual overcomes the illness effects (Robinson, 1990; Frank, 1995). The heroic accounts afford a clear progression towards values or life goals (Robinson, 1990; Soundy et al., 2013).

However, like the metaphor of a hero's journey Frank (1995) argued there is a risk in Quest stories. He expressed his reservations about metaphors such as the 'hero' or 'the phoenix' where people describe their illness experience as a 'complete transformation' (p.136). He opposed generalised metaphors offered as storylines as they can present the process as 'too heroic' and 'too clean' (p.136), thus depreciating those who fail to rise from their ashes. The quest story is not a goal toward something (Frank, 1998a) but rather an attempt to 'tell an illness story in one's own voice' (Frank, 1995, p.134). Ellingsen et al.'s (2021) study of the stories of nine individuals who experienced intensive care treatment, they exemplified narratives of quest by 'finding oneself after critical illness' (p.35). The stories encompass lessons people learned after experiencing critical illness that 'life could no longer be taken for granted' (p.40).

CHAOS

The Chaos storyline, lacking order, structure and coherence, can be hard to hear (Donnelly, 2021). Chaos is found in stories of the deeply ill, where disability increases with unremitting pain and the condition is unsuccessfully treated (Frank, 1998a). Chaos lacks narrative sequence and can only be described retrospectively (Frank, 1995). This storyline suggests that it is 'difficult' to listen to, as 'no one wants to hear' some parts (p.101), for it implies that no one is in control. To put it simply, chaos is life imagined that 'will never get any better'. Chaos is the anti-narrative of the dominant medical culture (Frank, 1998a), where restitution believes that 'illness can be cured', and a 'chaos story is devoid of effective action' (Frank 1998, p.202). Williams (1984) argued that some illness narratives lose 'the orderly sequence of facts' and 'cannot be sustained against chaos (p.178)'. Such that examples of chaotic storylines were exemplified in Nettleton's (2004, 2005) studies of people with Medically Unexplained Symptoms (MUS). With no pathological origins, MUS patients were unable to make sense of their illness and clinicians unable to provide therapeutic interventions (Nettleton et al., 2004). Similarly, Smith and Sparkes' (2004, 2005, 2011) studies on people with Spinal Cord Injuries (SCI) found chaos as a universal response, and people sought to impose order against impairment or disability. They suggest that 'chaos is no way to live', it is a story that 'one needs to move out of (Smith and Sparkes, 2011, p.45).'

However, Frank (1995) argued, 'the worst thing is to rush the person in a chaos story to move on' (p.110). Donelly (2021) observed that in western storytelling, chaos is usually 'erased or silenced' (p.3) as it does not fit the traditional format of resolution. This is particularly true in the stories of the chronically ill and the disabled, as they don't have a happy ending (Frank, 1995; Donnelly, 2021). Smith and Sparkes (2011) singled out a chaos story from a man who suffered an SCI injury. He recounts: 'Nothing to live for. It can only get worse. I may as well be dead' (p.41). The authors state that whenever they share his story with a new audience, they sense anxiety, discomfort, it instils fear and at times, the audience 'need[s] to respond' (ibid). Frank (1998) argues that the problem with chaos stories is that listeners must 'honour the telling of chaos while leaving a possibility of change', to accept what is told, 'without accepting its fatalism' (p.202). Frank (1998) advised to 'just listen' (p. 197), for to deny the chaos story is to 'deny the person of their experience' (p. 202). Moreover, Donnelly (2021) and Frank (1998) suggest that listeners should tolerate chaos stories, for narrating chaos is one way in which individuals 'claim agency and authentically voice their experience', in a world 'that censors and covertly prescribes' preferred illness narratives (Donelly 2021, p.13).

RESTITUTION

The Restitution storyline is the culturally preferred narrative as it emphasizes cure and the restoration of health (Frank, 1998a). It can be summarised as, 'Yesterday I was healthy, today I'm sick, tomorrow I'll be healthy again' (Frank 1995, p.77). Restitution views illness as transient and that the body can be fixed as 'good as new' (ibid). This can be connected to the Parson's concept of sick role (Varul, 2010), where the ill get treatment, they get better, and the future is restored. What can be heard is the powerful narrative of what medicine expects from the ill, and what social institutions expect from medicine (p.83). Restitution

celebrates the heroism of health professionals and medical science (Frank, 1998a), in which 'disease is the enemy' and cure is seen as 'conquering that enemy' (p.201). In Thomas-Maclean's (2004) study of 12 women with breast cancer, restitution was the most desired narrative, as it resembles 'what life might have been like' (p.165) had illness not intervened. In contrast, Whitehead's (2006) study of people with chronic fatigue syndrome/myalgic encephalomyelitis (CFS/ME) found that people with CFS/ME do not actively embrace a restitution narrative due to the 'constant reminder of the illness that they live with; the body is never symptom free' (p.224).

Restitution reflects contemporary medicine's focus on diagnosis, treatment, and cure. This projects a future that will not be 'disrupted by illness' (Frank 1995 p. 90). A linear trajectory of illness experience is expected; breakdowns can be fixed; the ending is a return to before the beginning: 'good as new or status quo ante' (ibid). This poses a problem that Frank (1998) identified as the social expectation of recovery, even to the point of demonizing illness. In my opinion, while in restitution it could be inspiring to hear stories of return to health, they may have an opposite effect for those that cannot recover; for 'they no longer work when the person is dying or when the impairment is chronic' (Frank 1995, p.94). Frank (1998) argued that the deeply ill are further marginalised as their 'reality does not include restitution', they also have no story that society considers worth telling' (p.201). When restitution does not happen, 'other stories have to be prepared' (Frank 1995 p.94).

MY OWN WAY TO PROCEED

My approach to analysis was largely informed by the work of Arthur Frank (1995, 2010, 2012), however I also periodically refer to the approaches of other narrative scholars including Reissman (2002, 2008), McCormack (2004) and Kim (2015). Frank (2010) believed that 'every narrative analysis needs to discover its own singular way to proceed' (p. 112). In this section, I present my own way of analysing and interpreting the study narrative data.

The data collection and analysis proceeded simultaneously. As Stake (1995) observed there is no particular moment when analysis begins, for 'analysis is the matter of giving meaning to first impressions as well as final compilations' (p.71). I observed that I was doing some form of analysis, such as noting my observations and summarising participants' accounts, in my field notes while conducting interviews. Consequently, after each interview was transcribed, I began the five analytical techniques used in this study (Figure 22). These were 1) The posture of indwelling 2) The interrogations of narratives 3) The acts of interpretation 4) The weaving of core stories and their return to participants and 5) The writing of an epilogue.

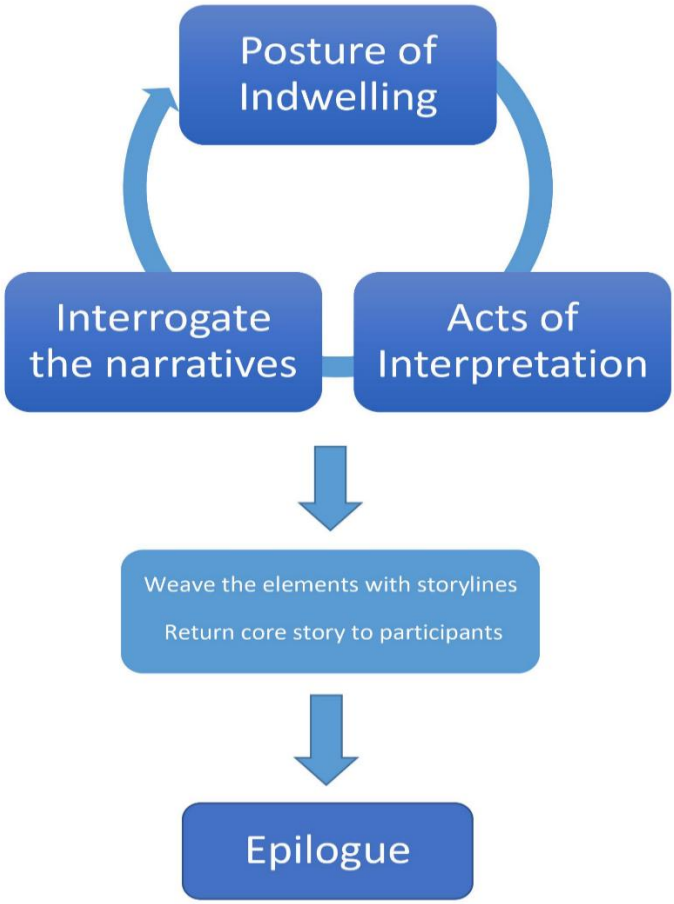


Figure 22. **The study analytical approach**

INHABIT A POSTURE OF INDWELLING

Firstly, I adopted what Sparkes and Smith (2012) referred to as a reflective posture of indwelling. This requires immersion in the data, including intensive reading and re-reading of transcripts. However, I did not limit myself to the transcripts, I also re-read my fieldnotes and referred to other data sources about the participants and our interaction. Riessman (2002) reminded me that analysis should not solely focus on transcripts but encompass the interview situation and the interaction.

The indwelling posture according to Maykut and Morehouse (cited in Sparkes and Smith 2012 p. 3), 'means to live within…understanding the person's point of view from an empathetic rather than sympathetic position'. I specifically dwell on the HD participants' personal meaning of their illness experience, rather than details relating to clinical symptoms. In the cases where family members and HCPs were involved, I explored their relationship with the person with HD and how their roles shaped the persons' illness experience. I would read each transcript, annotate, and subsequently refer back and forth to the relevant documents contained in the participant's narrative boxes. This helped me get a sense of the context and general feel of the participant's story.

INTERROGATE THE NARRATIVES

I devised a list of questions to interrogate the narrative data and to guide the analysis. This follows Frank's (2010) suggestion to use a method of questioning, for dialogical narrative analysis is 'not a prescribed method' with a set of steps to follow rather it is the 'practice of criticism' that involves 'analytic and interpretative movement of thought' (p.73-74). The following set of questions initiated the analysis and drew attention to the particular work that stories have the capacity to do, including 'to better understand the people you listen to or to make sense to the people you hope to listen to you' (Frank 2010 p.74). These questions were iteratively used to what was the most relevant to the data.

- What is the story they want to tell others and what meaning does it convey? (Kim 2015 p. 167)
- What does the story make narratable? (Frank 2010 p. 75)
- Why is someone choosing to tell their story, and what particular capacities does the storyteller seek to utilise? (Frank 2012 p.33)
- How does the story help people remember who they are? (Frank 2010 p.83)
- What multiple voices can be heard in any single speaker's voice; how do these voices merge, and when do they contest each other? (Frank 2012 p.33)
- Who is this person? (Frank 2004 p.434)
- How can the story be told to best represent his/her life? (Frank 2010 p.86)

My personal aim was not limited to the research question of, 'How do people with HD construct their illness experience?' but equally tied to Frank's (2002) questions, 'What sense shall I make of these stories and how shall I represent these to others?' (p.114). These interrogations resounded loudly as I was confronted with lengthy transcriptions, mounting documents including my own muddled, elaborated field notes and angst-ridden journal. My efforts to systematise the data in 'boxes' provided some kind of order, but I was frustrated with the rigid approaches and linear steps found in narrative literature. I finally understood what Frank (2010) meant by his instruction to 'let stories breathe' and to avoid the pitfalls of 'somebody else's rule' (p.2). Frank (2010) argued that the study of stories is 'more about the asking, what do stories do,' which ultimately 'inform us about human life' (p.2). This realisation has enabled me to adopt my own analytical strategy and make my own decisions around the study methods and analysis.

PERFORM THE ACTS OF INTERPRETATION

I used a set of practices that Frank (2010) calls 'acts of interpretation' (p.106) to aid with my interpretation of the data. Frank argued that interpretation is not about decoding stories but requires the researcher to open up and see all the variations and possibilities innate in a story. Interpretation is entering into an ongoing dialogue with a story, translating it and discovering the unnoticed aspects, such as the story's connections and discontinuities. Ollerenshaw and Creswell (2002) referred to a phase of narrative analysis where 're-storying' or 'retelling' occurs (p.329). Here, I used the following 'acts' to re-story and formulate the HD participants' core stories. Examples of how I used each act can be found in the Appendix L.

- Translate story into images (Figure 1, Appendix L)

I drew images based on what I recalled and understood of the content of the participants' stories. Frank (2010) advised the initial step of interpretation is to 'stop reading or listening and *see'* (p.108). The assembling of images helped me visualise the events that unfolded and how they connected to the narratives. This allowed me to evaluate my understanding of the stories, and in writing up the participant's core story, this made me aware of the imagery it evokes in the reader's imagination.

- Translate the story from another's point of view (Figure 2, Appendix L)

I wrote a summary of how the story could be perceived and understood by other people. For example, for each HD participant, I chose a family member they mentioned who was important to them and summarised how they see the person's story through their eyes. Frank (2010) and Riessman (2008) assert the dialogical nature of stories; emphasising that they are polyphonic and incorporate multiple perspectives. By translating the story into different viewpoints, this opened up new and different angles on the analysis.

- Appreciate the story and the storyteller (Figure 3, Figure 4, Appendix L)

I composed a total of eighteen letters corresponding to each of the nine participants and their narratives. They were handwritten but were never sent. The writing of letters although time consuming, has opened up my personal interpretation of their data. Furthermore, it helped me better understand the participant's identities and my relationship with them. In the letters, I wrote how the narratives personally affected me, and my appreciation of what I had learned from the participants. Here, I experienced what Frank (2010) meant when he wrote about the 'capacities of stories to do a particular kind of work' (p.74). These letters not only enlarged my perspective but enabled me to better appreciate the content of the story and the significant events of the narrative, as well as realising the personal effect of stories to me as a reader.

WEAVE THE ELEMENTS INTO A CORE STORY AND RETURN THEM TO PARTCIPANTS

I constructed a core story for each person with HD. This was done by weaving together all the elements, and the narratives that 'call out as needing to be written about' (Frank 2012 p. 43). Frank (2012) stated that which stories to select is crucial, and I made my selection based on '*phronesis*'. Frank (2012) described phronesis as 'a practical wisdom through analytical experience' (p.43) and argued that judgement on what to write is influenced by what the researcher has learned during fieldwork, and that often this knowledge remains tacit. Thomas (2010) posits that phronesis is provisional, and that it can only be tentative interpretation and analysis. I agree with Frank (2012) that phronesis is 'more of a craft and less of a method or procedure' (p.43) and with Thomas (2010) of depiction of phronesis as 'malleable and corrigible' (p.578). Writing the core stories was a lengthy process involving several drafts and constantly 'revising story selections as the writing develops its arguments and revising the writing as those stories require' (Frank 2012, p. 43).

Furthermore, I analysed the data in what Frank (1995, 2010) described as a narrative analysis rooted in a storyline. I weaved the data through 'the most general storyline that can be recognised underlying the plots and tensions of particular stories' (Frank 1995 p.75). *The Wounded Storyteller* (Frank, 1995), proposed that any distinctive illness story can be composed through a weave of at least three core narratives, namely Quest, Chaos and Restitution. I used these model storylines as 'guides to listening and storytelling' (Frank 2010 p.120). The identification of the storylines was 'sufficient to structure a report' (Frank 2012 p.46), and rather than generalising people's experiences, the storylines recognised the uniqueness of the person's story at the same time as acknowledging that an illness experience can draw upon the availability of narrative resources.

These storylines were not easy to recognise. Like Frank (2010), I felt it was hard enough to analyse the content, but also that I had to analyse how participants dealt with problems of narration, particularly how they construct their narratives. From the standpoint of a storyteller (Frank, 2000), it is clear that we humans live a multi-layered contextual life. As I engaged in the study process and knowledge generation, I uncovered the intricate relationships between our experiences, social worlds, and identity. The stories we tell about our lives are messy and multifaceted, and the study demonstrated the usefulness of storylines to help navigate and sort out the complex narrative threads that constitute a story. Writing the core stories was the most challenging and time-consuming part of the study, not only in making decisions on what to include, but at the same time I was conscious of how the participants would respond when I returned to them these enriched stories and my interpretation of their experiences.

The core stories were first shared with my supervisors for feedback before sending them to the participants by either email or post. They were accompanied by a letter asking them to respond to a set of questions (McCormack, 2004), as seen in Table 13. The core stories were co-constructed, and I wrote the final versions after consulting with the participants and asking for approval.

Table 13. Core story questions, adapted from McCormack (2004)

Core story questions
Does what I have written make sense to you?
How does this account compare with your experience?
Have any aspects of your experience been omitted? Please include these wherever you feel it is appropriate.
Do you wish to remove any aspect(s) of your experience from the story?
Please feel free to make any other comments.

WRITE THE EPILOGUE

I reflected on the feedback given by the participants after they read their core stories, and subsequently wrote an epilogue. McCormack (2004) suggested that an epilogue allows 'reflections on the stories' and to

'close the narrative' (p.221). However, in contrast to McCormack (2004), who mainly relied on interview transcripts to construct the epilogue, I used all the available narrative data, including the reflections written in my journal. However, the study's longitudinal approach with multiple interviews and multi-sourced data would mean that the coda of each HD story could only be written when all data had been collected and after the exit interviews had been conducted. The epilogue concluded the study analytical strategy.

SUMMARY

This chapter presented the design, methods, ethical issues, sample, and analytical approach used in this study. By conducting this type of research, it extends our knowledge and understanding of the HD illness experience by pursuing new avenues of inquiry and creating an in-depth, focused, and contextual study centred on the person with HD. The use of multiple narrative methods of data sources tailored to the person with HD conducted in a longitudinal approach has previously not been explored in HD qualitative studies (Audulv, Packer and Versnel, 2014; Mahmood, Law and Bombard, 2021). Narrative methodology is particularly important in studying chronic illnesses (Charmaz, 2002), life- limiting (Bingley et al., 2008) and genetic diseases (Petersen, 2006), where the voice of the person living with the illness, such as HD may be particularly hard to hear (Audulv, Packer and Versnel, 2014). Frank (2009) and Reissman (2008) maintain that the study of narratives begins with putting a human face to a story for people who need to hear them. Reisman (2002) states that,

Narrative methods *'are not appropriate for studies of large numbers of nameless, faceless subjects. The approach is slow and painstaking, requiring attention to subtlety: nuances of speech, the organisation of a response, relations between researcher and subject, social and historical context. It is not suitable for investigators who seek a clear and unobstructed view of subject's lives, and the analytical detail required may seem excessive to those who orient language as a transparent medium' (p. 706).*

In the next chapter I present the three stories of people with HD, namely Jen Walters, Martin Taylor, and Daniel Carragher.

CHAPTER 5: THE HUNTINGTON'S DISEASE STORIES

'I want to understand the world from your point of view. I want to know what you know in the way you know it. I want to understand the meaning of your experience, to walk in your shoes, to feel things as you feel them, to explain things as you explain them'. (Spradley, 1979 p.34)

This chapter presents the study findings found in the stories centred on three people with Huntington's Disease (HD), namely, Jen Walters, Martin Taylor, and Daniel Carragher. All have manifest HD symptoms, and are respectively in the early, middle, and late stage of the disease. Whilst this study was mainly influenced by the work of Frank (1995, 2010), I was also inspired by the works of clinicians such as Kleinman (Kleinman, 1988) on chronic illness and Bird (Bird, 2019) on HD. Drawing from patient encounters in their clinical practice, Kleinman and Bird acknowledged the capacities of stories to provide personal meaning for those who are ill and all those who care for them.

Each narrative case was structured with the introduction of the person with HD in the context of the research setting. I proceed with their story which features the three storylines of Quest, Chaos and Resignation based on my analysis of the research data. The storylines helped structure the narratives in presentation and form (Frank 2010). The story culminates with an epilogue based on my reflection of our final research interaction.

The stories emerged through collaboration with the participants during 18 months of the study. They were actively involved in the narrative construction by being invited to make comments on and edit the transcripts, and to analyse their core story. Like Kleinman (Kleinman, 1988), I made extensive use of verbatim quotations obtained through direct transcriptions of the interviews. For ease of reading, where quotes have been used, 'narrative smoothing' (Kim, 2015, p.192) has been employed, I omitted the '*erms*', '*uhhms'*, and other speech sounds and redundancies, except where they were deemed important to keep the meaning of the text.

At the core of each story is the illness experience of the person with HD. The first story starts with Jen Walter, at her late stage followed by Martin Taylor, in the middle stage, rounded off by Daniel Carragher, in his early HD stage.

JEN WALTERS

She was staring at the screen. From my laptop window I could see her in a wheelchair, her hair long, unruly, and grey. She sighed, incomprehensible words that didn't begin or end. She gave me a smile, then clearly said; '*Thank you Paul, come visit me if you have time.'* Her right arm attempted to reach my face, only to be reminded we are in a virtual space. Her carer, whom I couldn't see but could hear her faint voice, reassured her, '*It's okay Jen, I'm holding your iPad'.* The video call ended.

It was in February 2020 when I visited Jen Walters in Driftwood Nursing Home for our first research interview. She stood at 5'5" and weighed about 120 lbs. She was wearing a Breton shirt and navy trousers. The black nautical stripes and her cropped dark hair reminded me of those sophisticated French ladies in movies. Her frame, like a graceful bamboo, swayed in different directions and her arms failing and legs shuddering. '*I can't walk much'* she said. She treaded from the lounge to her bedroom, about two metres, and plunged into her armchair. She pointed at her bed for me to sit down. I remembered Jen holding a plastic water container in her hand; it resembled a sports water bottle with a flip straw in it. Jen was drinking from it periodically leaving stains on her top. That was the last time I saw Jen independently drinking and walking.

Jen Walters is a 56-year-old Caucasian woman who has been living in the nursing home for the past five years. Born into a family on the south coast of England, Jen has another sibling who was been recently diagnosed with HD. Her family suspected that the inheritance of the HD gene might have come from her father, however this was never an official diagnosis. Jen's father was diagnosed with MND and passed away. When Jen received her HD diagnosis in 2011, *'I was in shock. I didn't know that HD exists in the family. I think my Dad had it. HD is a family thing, isn't it?'* Jen further stated, '*I am happy living here. I hardly think of HD now'.*

Jen used to teach English in a secondary school for half a decade. A job which in her words *'I enjoyed doing'*. Jen was never married, she had three children. Her eldest son, Timothy in his 30's was born out of her relationship with her first partner, and from the second partner she had another two children, now both in their mid-20's: a son, Francis, and a daughter, Marjorie. When I asked about the fathers of her children, she turned her head away. I gave her time only to be met with further silence. I gleaned from her gesture the need for privacy. However, when I asked about her children she stood up, placed her water bottle on the bedside table and sat closer to me. She returned to the conversation, eagerly sharing aspects of her life again.

Jen described her daily routine in the nursing home as *'enjoy what I can'*. She would wake up around 8 in the morning. The morning shift of carers will help her to have a shower and prepare her breakfast. *'I try to join in with everyone* 'sitting in the common lounge or engaging in the activities in the nursing home. Jen enjoyed visiting the museums and local attractions. Her love of art is reflected in the colourful paintings,

and art prints in her room. We spoke about her books nestled in a tall bookcase, from the classic works of Jane Austen to the contemporary philosopher Alain de Botton. She fondly talked about her previous pets, her cats Betty and Charlie. Consequently, she showed me two cat paws tattooed on her arm.

Jen's day usually ends with an evening meal at 5pm and the rest of the day she would stay in her room. I asked her what does she do at night?, and she said, *'It could be anything. I usually sit in bed and listen to the music. I have supper then I go to bed.'* Jen's evenings are mostly spent in her own company. She showed me her radio antenna and how she liked to tune in to BBC Radio 4. She haltingly said, *'I couldn't do much now…'*

JEN WALTERS AND ME

I used to be Jen's physiotherapist for two years. I knew her from a patient-clinician relationship. Occasionally we would talk about her plans for the day or things that happened since her last visit. The majority of our conversations revolved around her mobility exercises.

Before I left clinical practice to pursue a full time PhD, Jen gave me a card which said*: 'I will miss you and good luck.'* I told her that my PhD research would involve talking to people with HD and she said if there's anything she could contribute to let her know. I bore that in mind.

I had previously met Jen's children in the nursing home. I would greet them in passing if we crossed paths in the communal areas. The eldest son Timothy, I had the most interaction with. Timothy is Jen's next of kin (NOK) and has been appointed with lasting powers of attorney (LPA). NOK means the closest living relative. LPA means Timothy has the legal power to make decisions on Jen's behalf if she is unable or lacks the capacity to make decisions for herself on issues around her health and welfare. Timothy and I occasionally found ourselves in a meeting room with Jen and other health professionals. Our conversations were centred on aspects of Jen's clinical care.

In late 2019, I sent a letter to Jen to see if she would like to participate in the research. The participant information sheets (Appendix I) were sent through to the nursing home, along with the advice to share the research information with her nurse and NOK. Consequently, Timothy sent me an email stating that Jen had expressed an interest. I spoke to Timothy over the phone, and he updated me that Jen's HD progression has impacted her physical abilities and her speech was increasingly slurred. I accordingly sent an adapted participant information and consent form (Appendix H) through the post which included reader-friendly information about the study. Once the forms were signed and completed, I scheduled a visit to see her.

On the day of our first interview, I was greeted by some of the nursing home staff who knew me. I had to explain to them that I am not visiting as physiotherapist but as a researcher. I wanted to be clear of my position and my purpose. I went to speak to the nurse Maria Crawley, to inform her of the reason for my visit. Maria gave me a handover there were no other safety or health concerns regarding Jen's

participation. I saw Jen sitting in the lounge, there were sofas and chairs facing the big television mounted on a wall. The musical film Grease was on TV. There were other residents and carers in the room. Jen was sitting on a two-seater beige couch next to a carer, and rather than facing the television she was facing the door. After a few attempts at getting up from her seat, she hastily stood up as soon as she saw me.

Jen said she would like to talk in her room. I asked her if she understood the purpose of the research, what her participation entails and if she had any further questions. Jen showed me her signed copy of the participant forms. She had placed it in a brown paper envelope which she kept it in drawer. Jen said she was pleased with everything and continued saying, 'Thank you.'

Our plan for more interviews and non-participant observations didn't materialise. The Covid-19 pandemic hit globally, and the national lockdown was imposed a month after I visited Jen. Non-essential visits were not allowed in care homes across the country until all restrictions were lifted in England in March 2022. The rest of my research interaction with Jen was through remote interviews of telephone and video calls, until we met in person again for the exit interview.

THE CONSTRUCTION OF JEN'S STORY

Jen participated in a total of six research interviews over the 18 months of the study. Her HD symptoms manifested the advanced stages, requiring full-time nursing care. Despite the progression of her disease symptoms, Jen's understanding of the research study remained generally intact. Her linguistic abilities deteriorated before her comprehension which is common in HD progression. Throughout the study, Jen demonstrated informed choice, not only in her daily activities but also with complicated decisions over her care. This was exemplified by giving consent on having a percutaneous endoscopic gastrostomy (PEG) operation in Sept 2020. As her nurse, Maria Crowley would put it, *'She is still up there'*. However, in our interactions Jen preferred short interviews (average 30 mins) traversed in frequent sessions. This was probably due to her reduced attention span, inarticulateness in speech, slowed thinking and difficulty in following complex instructions associated with HD late stages. I particularly noticed Jen's physical restlessness with increased gross movements when we talked over long periods. To overcome this, I offered Jen sufficient intervals between questions and promoted personalised varied methods of communication. Drawing from HD studies on the effectiveness of low technology communication aids (Ferm et al., 2010; Diehl and Riesthal, 2019; Grimstvedt et al., 2021), I utilised pictures and objects of reference, as well as making use of Jen's personal mementos to maintain our discussion. Not only did this give her a sense of 'communicative control', but it also maintained her interest in the study. These strategies appeared to help with her thought processes and facilitated autonomy with her answers by using resources available to her.

Jen's story was further illuminated by the accounts of her children, Timothy and Marjorie Walters, her social worker, Poppy Williams and nurse, Maria Crowley. This organic interaction came about when Jen suggested I talk to Timothy. After the interview with Timothy, he suggested that it would be beneficial to

talk to his younger sister and the social worker particularly at the time when he was living in another city whilst studying in the university. In total there were five individuals involved in Jen's case. For Timothy, not only would this fill the gaps in the narrated events but also will *'shed some light on some different aspects'* of the disease. From the perspectives of Jen's children, her participation gave her *'a sense of purpose'*, '*that for once she could just be herself*' to share her story.

THE PORTRAIT OF A DISEASE

PART ONE: QUEST

IN SEARCH OF ANSWERS

Jen Walters now hardly thinks about HD, in stark contrast to the time before her HD diagnosis. Jen had spent more than 10 years needing to know what was wrong with her. She suffered from severe depression. In her 40's she was trying to understand her hasty clumsiness and difficulty completing tasks. At the time she found consolation when doctors told her she had dyspraxia and attention deficit hyperactivity disorder (ADHD). She reached out to people with similar conditions in online groups, but soon her mental health worsened. Although the prescription drugs and the cognitive behavioural therapy (CBT) briefly eased her anxiety, the amalgam of diverse symptoms only added to the confounding diagnoses.

Timothy remembered, *"When I saw my mum when I came back from university having not seen her for a few months and it kind of being 'oh, you're looking a bit wobbly' you know."* Jen became increasingly unsteady on her feet, which led to frequent falls at home. After several visits to her neurologist in 2011, the genetic test confirmed that she had HD and, a little later, Jen passed this information to her children. The HD diagnosis came as surprise that impelled everyone in the family in the search for more answers.

Marjorie Walters recalled, '*My mum was using my laptop to send e-mails, I was about 13 and she wasn't very good at being tech savvy or anything. She struggled a bit with sending e-mails and whatever, so she left her e-mails open on my laptop and she had sent an e-mail to her friend to tell them that she had received confirmation that she has Huntington's, and she needs to get rid of the car. And that was the first time I'd ever seen the word Huntington's. I had no idea what it was.*'

A quick Google search of 'Huntington's Disease' yielded over 4.6 million results (June 2021). Marjorie's search in Wikipedia imprinted on her mind the images of the brain and the word 'dementia' stood out. For Timothy, he remembered his grandad *'becoming unwell, falling over with this crazy gait'.* The Walters' family search for the definition of the disease led to different inferences and meanings. For Jen, it was the incurable hereditary progression. Jen was trying to come to terms with losing her capabilities, but her primary concerns were that *'I worry most for the kids'*.

Being a teenager at the time, Marjorie found; *'it was a bit too overwhelming, I didn't really know how to process it'.'* For Timothy,*' there was a lot of kind of learning to do very quickly and I didn't know what HD was at the time. So, you know, I had a crash course and at that point it became very real.'*

Reality set in when Jen got rid of the family car. Living in a secluded part of the town, giving up driving was a gesture of Jen relinquishing her independence. Soon after, she accepted assistance at home when community carers came to support her day-to-day activities. A personal assistant named Julia Broderick, recruited by Jen's mother, came regularly to assist with shopping, cleaning, and other household chores. Despite describing Julia's presence as *'being helpful'* and confirming that she had a *'great relationship'* with Jen, Timothy recounted, *'This is something that was always really difficult. Because you have to admit to yourself and to everyone that you need help. So, I remember that being quite a difficult thing for mum'.*

SOMETHING AMISS

Timothy and Marjorie distinctively remembered growing up in a house which was '*constantly a shit tip'* , *'full of to-do lists everywhere'.* They were cognisant of Jen's behavioural volatility, ranging between periods of depression and aggression. The revelation of their mum's HD diagnosis explained what was happening in the household.

Marjorie said, *'The house was very chaotic, and she'd gradually drop things more and it was just, you could tell that there was something not right.'*

Timothy recalled, *'A constant mess, 'Is there a bit more to this?'. I remember thinking mum is really struggling with cleaning. All these kinds of little things creeping in adding to a general sense that something is not right. Also, being aware that mum's mood is becoming more and more unstable.'*

Marjorie remembered her mother's volatility, '*Well I definitely noticed mum's bursts of rage every now and then, which I never understood what they were but obviously now looking back I'm so much more understanding because it wasn't her fault. She would sometimes have this outburst of rage, but it wasn't her. Afterwards she would say 'oh I'm sorry Marjorie, I'm being really crabby' and you know, we'd get over it. I remember, before my brother and I went to school, it has just stood in my memory, she was screeching at us out the door and then we left to go to school and I just think I remember being like 'this is just so not, something's just not okay.'*

Throughout my conversations with Jen, she described how these times *'were not good'* as she was losing control of things around her personal and family life. She was angry that she was previously misdiagnosed and frustrated with her diminishing capabilities. Marjorie and Timothy recalled the many instances where their mum was close to taking her own life. Timothy added, *'I think this period in particular is a really difficult one and it's that period where the sufferer is still independent, they're still lucid, they're still, you know, can go about their daily routine to a point but they are losing grip on themselves and everything around them and it starts to manifest in these really subtle but very damaging situations'.*

The damaging situations that Timothy referred to were extended to public places. On one occasion, Jen was turned away in a café and accused of being drunk because of her reeling gait. She became irascible and her uncontrollable vexation made many social situations worse. *'Loads of people would look at her, it was upsetting for her, mostly for her, it was upsetting for me as well',* Timothy added. Jen felt increasingly isolated as people were quick to judge her uncontrollable body movements. Gradually Jen withdrew from social situations and turned to alcohol. Her justification, like that of many other before her was to cope with stress which consequently made her HD symptoms worse. A couple of cans of Stella beer turned to an evening routine multiplying in number, increasingly incalculable over a year. Marjorie recalled Jen insisting, *'that we had to stop at the shop to get it. Even if we said 'no', she was 'we have to'.* The sombre nights that Timothy remembered seeing his mum *with 'tins of beer and she'd just sit on the sofa and drink, and she became really apathetic.'*

Marjorie became so used to seeing her mum sitting on the sofa, drinking *'It was horrible, absolutely horrible. Mum had no desire to go out, no desire to live, just not really living at all, just completely existing. Really quite depressing.'*

THIS BECAME TOO FAMILIAR

The deterioration of Jen's mental health manifested in her increasing suspicion of the people around her. Jen distrusted her personal assistant, Julia, and accused her of stealing. She stopped talking to her best friend, Samantha Harris, who was helping with her finances. Jen cut ties with her mum. She believed that they were all in a conspiracy and were not acting in her best interest. Jen's paranoia, delusions, and aggression came with the potent mix of her alcohol dependency. Jen soon turned against the people who supported her. Inevitably, she disengaged with her doctors and other health professionals.

Marjorie added, '*She pushed them away, she didn't want to speak to them. She knew that she was losing lots of her abilities and she was panicking about, you know, the worst-case scenario. I think why she was worrying about it is because she was worried she wouldn't be able to do anything and so her way of gaining a bit of control was to just try and cut them out and to keep herself safe. Obviously, a lot of it is completely, it's just nonsense but I think it must be a bit of a coping mechanism that happened.'*

Timothy reflected on how his mum struggled with daily tasks: '*their mood is very low, they're struggling to communicate and they're having to juggle various appointments with neurologists and physiotherapists, and they've got you know, especially, money is an issue as well. They're having to juggle trying to get a social worker. They have a social worker that isn't being particularly helpful and all this kind of stuff. I think it's quite easy for them to decide that everyone's against them.'*

These were the all too familiar things that were happening before Marjorie's eyes when her role shifted from a daughter to her mum's custodian. *'I was living with mum and I was so, you know, I was an extension of her really so I found it quite difficult. Gradually things were getting worse and worse and I was living with mum so the changes that were present would sort of be so familiar to me that I almost couldn't really see*

them'. Jen's increasing alcohol intake had become so problematic that it made the home environment unsettling. *'There was no real way of stopping it, it was just really difficult to, once she had her mind set on something it kind of just, that was it, you know. Yeah, it's really difficult to explain.'*

Marjorie added, *'I just gradually noticed that I was actually her carer, but I don't know when the shift happened. It just kind of happened and I remember just one day being, like, okay, you know, some things I'm definitely responsible for my mum now. '*

PART TWO: CHAOS

THE CRISIS AT HOME

Jen turning to alcohol was the start, according to Marjorie and Timothy, of the '*dreadful times'*, the *'dark path of mum's journey'.* Jen's hallucinations and delusions persisted. Her aggression moved from the purely verbal to the physical, and in one incident she pushed a visiting carer down the stairs. She refused her medications. Her delusions intensified and she began making accusations against people, such as her doctor, who she said was a sex-offender. She was suspicious of her neighbours, some of whom she accused of maleficence. She talked about imaginary people in their street and posted inexplicable letters in random letter boxes. It was *'upsetting and disturbing'* to Timothy that Jen had become a risk to herself and others because of her irrational behaviour and paranoia. She was no longer safe at home, and consequently Jen was sectioned under the Mental Health Act and admitted to a psychiatric hospital.

Whilst under the care of the community mental health team, Jen was periodically confined in psychiatric units and care respites. There was a short period when Jen was discharged back at home, whilst a plan for a support package was put in place. The plan included home improvements such as disability access, given the deterioration in Jen's mobility. Before this was implemented, Jen's alcohol dependency and psychiatric symptoms worsened. Marjorie recalled, *'There was no sense of urgency and it was too late'*. Jen was once more detained in the psychiatric hospital, according to her children, because the health system had been unable to help them, and health professionals had given up on her.

Timothy felt helpless as the care professionals were not fully understanding of Jen's condition. *'They set a care package up, the care package failed, she'd get sectioned again and I think she was sectioned about three times over the course of that 18 months."* He expressed the frustration that *''it was quite naive and obviously there was a lack, a general lack of understanding of the condition. They'd say 'you know, your mum is refusing to take her medicine so she's putting herself at risk', well, but not understanding that actually it's, that's not her choosing to do that, it's, you know, it's, you know and they'd say, 'we can't deal with her because she's insisting on, on lighting her cigarette on the stove, she's gonna set her hair on fire and even though we keep telling her not to she keeps on doing it' .*

Marjorie remembered this time with sadness, *'I think the fact that she ended up being quite so isolated with no support and just nothing and there was no-one there to sort of, you know that act of finding people*

or finding the right advice, she couldn't do that. We also couldn't really do that because, like, no-one seemed to be, like, particularly supportive or actually, it just seemed to be such a chore and I think the fact that I couldn't, how the hell is someone with Huntington's going to when that kind of task is just so hard and it was really hard.'

'I feel like that sense of freedom and autonomy was just ripped from her and I completely, you know, it was the most painful thing to watch, and I completely understand her. I'm not saying that it was okay but her use of, like, drinking and this delusional, you know, I understand, like, where that's come from because all of her abilities were just being slowly sort of taken from her and she didn't really have a quality of life so what do you expect?' Obviously you're going to probably deal with it in, in whichever way you can and that was her way of dealing with it but it was just annoying at the time that people would see the alcoholism and be, like, 'oh well, I can't help you because of this' or whatever, there would always be an excuse as to why, it was sort of my mum being an alcoholic or whatever rather than someone living with Huntington's and not having the support and it was really hard.'

Marjorie observed that *'No-one there cared about her, it was really horrible.'* She said*, 'It was at this point where we were like well she was there for a long time because we were having to set up a 'best interest' meeting, you know, and all of this kind of stuff had to happen in the background and make the huge decisions to essentially take away her liberty and decide where she was gonna live. So, all of this stuff had to happen while she was kind of on her own in this and it's really upsetting.'*

THE BURDEN OF A MOTHER AND HER CHILDREN

In Jen's words, '*I was very sick in the hospital'.* It wasn't clear if she was aware of the specific incidents that occurred at the time as vividly described by Timothy and Marjorie. Further, Jen mentioned a *'drinking habit'* which she didn't elaborate. She recalled that she had been in different care institutions but couldn't remember for how long for, the details of her experiences, or the reasons for admission. Jen's memory gaps coincide with the time of her violent episodes and before her diagnosis of HD induced psychosis.

In my many conversations with Jen, she highlighted the period of receiving her HD diagnosis and her current experiences in the nursing home. In between those phases was a blur. When I pressed her on the 18 months in question, Jen repeatedly answered, *'I couldn't remember much'.* Sitek et al (2014) suggest that while memory impairment can be associated by HD progression, denial can be a putative coping mechanism. Whether this was a void in her memory, or she didn't want to talk about what happened, only Jen can answer. But one thing is certain, those were the difficult times that caused innumerable emotional burdens for Jen and her children.

Recalling the past revealed a profound sense of guilt for Timothy. He went to university and lived with his dad at the time his mum started becoming unwell, *'This was something that was in the back of my mind, and something that was beginning to occasionally kind of upset me and I was worried for her, and I was.'* Timothy found himself helpless to improve his mother's situation. He was torn between living his own life

or going back to look after his mother and younger siblings. *'I was really anxious about my mum's mental health and her mental wellbeing, and I knew she was so low, and I just wanted, I, it was really, that was the thing that upset me the most at the time.'*

The turning point for Timothy was when he received a phone call from Marjorie when 'she *didn't know what to do'* after Jen's refusal to let anyone in the house. *'The whole situation just imploded. I met up with Julia, away from the house, and she was just, like, 'Look Tim, things are getting really bad, your mum's talking to herself, she's very paranoid' and basically it became quite, it became apparent that mum was reaching a point of complete mental breakdown and, and in hindsight and having spoken with different doctors, you know, what happened over the ensuing kind of months is mum went into Huntington's induced psychosis and she completely, I think her brain and her mind and her, you know, just her kind of, she couldn't take it anymore and she just, erm, she just completely snapped.'*

This had a profound impact on Timothy because he acknowledged that his younger sister was having a very difficult time and that he needed to step up and get involved in his mum's care: *'I wasn't at home for, I wasn't at home for the really bad parts.'*

Marjorie, the youngest of all the siblings carried the load of looking after her failing mother, and during our conversations, I felt her sadness and indignation. As Marjorie said, *'It was just myself and my mum for quite a, well the majority of my childhood. I mean I think I was trying to juggle my education, whether it be secondary school, college etc, and you know, having a social life, which at that age is the most important thing ever, and then kind of, coming home to this really, really different environment which I just, I couldn't really comprehend, I couldn't comprehend the difference between my, you know, being at home and then being out with my friends where they just didn't have anything to worry about.'*

The situation took a toll on Marjorie: *"Yeah I mean, I don't think I knew at the time what was going on in my mind, I was just, erm, I went into this really weird, I remember it happening, I sort of got so overwhelmed with everything and then I feel like I just went into autopilot one day and I just need to, like, I can't, the more that I think that, like, you know, I can't actually cope with it if I sit and think about everything so I need to carry on, like, survival mode because it was all so surreal. I just hadn't been prepared for the, the, the depth of what I was meant to deal with."*

She added, *'Obviously my own emotional maturity and kind of genuine, like, you know, I was still a teenager so I actually think it was incredibly, I think I will always be quite upset that it was me, like, a teenager that had to deal with it because I also think I probably, knowing me, can a teenager really deal with that kind of thing properly? Like obviously I did the best that I could, I really did but I also think how unfair for mum that that was who, I don't know. It was a really tricky situation. I think I was just trying to do the best that I could really, and I had so many different things to juggle.'*

As Timothy felt the guilt, Marjorie looked back with resentment recalling the time at home where Jen sustained a fracture and needed to get emergency services, *'I had this bit of, slight bitterness at the sort of*

amount of responsibility that had to fall on my shoulders without, you know, any, it just happened and I'm not, obviously I'm so glad that my mum was okay but, I think I had a bit of anger towards the situation.'

Marjorie needed resolution and shared with me her thoughts on growing up in the shadow of her mum's HD. '*I think my sense of self, my genuine identity was a complete mess, like, I had no idea who I was because whilst everyone else was sort of developing relationships, friendships, whatever I was just genuinely at home trying to make sure my mum didn't, you know, cos she would avidly smoke all the time and I was just worried that she might burn the house down or fall over and, I was constantly worried. I developed anxiety, of course, [laugh] cos it was just, you know, consistently, erm, worrying about, I think when you live in that state where something bad could happen at any point how can you have, how can you not be anxious, it's just, absolutely crazy and I think I personally had a bit of, I just felt rather isolated from, as much as I love my family and [laugh] you know, I think I needed someone else to kind of step in and be, like, you know 'this is way out of your depth', but it just carried on for such a long time.'*

"I was as much of a victim, not as much of a victim but I was definitely a victim in that situation and I'm not saying mum, I just think HD, like, you know, caught me and I was sort of really not prepared, erm, but I just, yeah, I think, I don't think people really understood what to do or how to help me and I don't think I really understood how, I don't think I really understood what was going on at the time or how to be, get help."

OTHER FIGURES

Marjorie and Timothy reached out to their biological fathers. While Marjorie didn't find comfort in her father's response, Timothy found his dad was his source of support. They both acknowledged that they needed to turn to their friends and family for emotional support. Marjorie found comfort from the advice given by an HDA youth worker. Although the presence of an HDA advisor was very helpful, she felt that their time was constrained. Throughout this period, it was this scant social support that became their lifeline. Jen's children had to fend for themselves and rely on their personal attributes to cope with the situation. Eventually, Timothy and Marjorie received counselling.

Timothy remembered, "*I had, like, maybe six weeks of counselling, and this was, this was during, when mum was in and out of, when mum was kind of stuck on the psych ward. I think I remember it being during that point because yeah and it's probably a good thing. I did that because I had to do a lot of, erm, I had to do a lot of crying just cos I felt, I felt really, I was just overwhelmed with guilt to be honest, that was the biggest thing. I just felt, I felt guilty that she was on her own and just stuck on this horrible unit in a, you know, with no-one around and just completely isolated and I just felt, I felt utterly dreadful because of that and I, I felt like I, even though I know that I was doing all I could I just felt like I wasn't doing enough or, you know, I wasn't expediting it as quick as it could have been, you know. '*

Marjorie recalled that, *"I got a bit of counselling when I was in when I was in college, yes, and she was helpful in giving me another perspective of what was happening. I think for a long time I didn't really, I kind of just sort of, I just had to kind of plough through, but then eventually my, well, I mean I tried, I think I tried*

to reach out a bit to my dad but I don't think it really went anywhere because he didn't really know how to deal with it. I eventually got this sort of crazy amount of help from my mum's friend Samantha Harris, but this is a bit further along the line, along the line but she kind of ended up stepping in and taking me under her wing and she, I basically, I think of her as a second mother really because she's basically been this loving figure in my life since, I just can't explain, I obviously wish that she'd been there for a lot longer but I'm so grateful that she came along cos she completely changed, she completely changed my life. But at the, before that I don't really know, I think I would try and get my head down and stick, like, I'd do all of my college work and kind of use it as a bit of an escape, so I did well, I did well at college actually and I think I actually used that as my coping mechanism which probably sounds a bit weird, erm, but, yeah."

'SHE IS AT RISK OF DYING'

Jen's children felt helpless while their mum was in and out of the psychiatric hospital. They viewed the health professionals who were involved in their mum's care as unhelpful and not fully understanding of her condition.

Marjorie expressed her frustration, *'You know, you need someone to know, like, what's actually happening cos you can't begin to explain it if they don't have a general understanding and I think most of the healthcare professionals were just very dismissive and just saw, you know, they'd only see mum's alcoholism or the delusions and they'd be 'oh she's this' or 'she's that' rather than being like 'well what, ... 'you know, look at this holistically because this is a product of being or living with this debilitating illness and not having the right support in place…'*

The interaction of Timothy and Marjorie with the health services resulted in dejection. They felt most health professionals saw their mum as an alcoholic, aggressive woman who refused to engage with them. They failed to see the person behind the HD, who was their struggling mother with no control of her disease.

Of the many scenarios that Timothy evoked, *'I remember getting infuriated because I could always hear 'well it's your mum, your mum is putting herself at risk' you know, like insinuating that she is aware of the risk that she's putting herself in in certain situations and all this kind of stuff. And it got to a point where, erm, what happened, I think I came down, it was one of my trips down and, I was I think there was, I think one of the nurses was there and mum was just refusing to engage with anybody. She would often do that at this point, she was getting so upset and frustrated with everyone around her she would just refuse to engage and just shut down.'*

'I have a memory of sitting in the car outside with one of the care team and just being in floods of tears and we were on the phone to our social worker, and we were just, like, 'look, this is a joke, she needs more than this' you know, and it was just, like, I felt utterly helpless to be honest. I didn't feel like there was anyone I could talk to, I didn't, I felt like, I think this is probably a key point actually is that I didn't feel like, her social worker who really should have been the person that I could go to and be like, 'look, what can we do here

because this is just, everything is going wrong and my mum's getting worse and she's, she's not being ...' and I just felt like she didn't get it really, like it was an inconvenience actually.'

Jen became *'a shell of a person'* in institutionalised care. Marjorie found her mum repeatedly sedated due to her behavioural outbursts and her non-compliance with care interventions. These were traumatic experiences for her children. Jen Walters eventually stopped eating, so much so that she *'became thin and poorly'* and *'she was at risk of dying'.* Marjorie thought what was happening to her mum was surreal. She recounted despondent images of her mum *'on a hospital drip'* and recalled a time when *'she had been sedated with something and, went to visit her and it was really quiet, it just felt like she was an animal.'*

'PERHAPS, THIS NEEDED TO HAPPEN'

While Jen was detained under the Mental Health Act her physical health was also rapidly deteriorating. The 'best interest' meetings continued, and her case was escalated to a 'crisis'. Consequently, a new social worker was allocated, Poppy Williams, who had over 10 years of experience in social care with elderly dementia patients. Poppy found Jen Walters' case unnerving: *'I'll be honest with you, my initial feelings and emotions were I have no experience of working with someone with this condition and there was that kind of natural fear, I think, as a professional when it's something that you're not used to working with and then because my background is in older people's mental health so to work with a family where two of the children were still in education, one was at university but the daughter was doing her GCSEs, was quite daunting for me because that was a dynamic that I also wasn't really used to working.'*

Poppy found herself at a loss, like other health professionals, as Jen disengaged with her. She sought advice from the HDA advisor, who drew her attention to much helpful information. In common with Marjorie, she found the charity's work very valuable, but they were stretched and limited in their capacities. Jen Walter's case consumed her, given the complexity of the symptoms and the family dynamics involved. There were many hurdles to overcome but one thing that Poppy promised Jen's children was to *'never give up on them.'*

For Poppy it was a difficult and lonely experience, *"When I had that case it would be the thing I would think of when I got up in the morning, it would be the thing that I would think of when I was driving home, you know.*

'I could see the injustice as well and I'm not gonna lie, it was really hard, when you're working with a person that won't even engage with you. At times, she has been quite aggressive to professionals and people, but it was about understanding that she's a person and the person behind that, you know, was an amazing lady that's going through something that's so horrific and the easy thing would be to stand back and go 'well this is all we can do' but that's not right is it?'

'I'm not someone that gives up easily. I will always, always try and do my best to help people and I think the moment I don't care I know I shouldn't be doing this job. I do think it's a weakness of mine sometimes, I

do think I care too much, that's probably why it consumes me. And I think just, erm, you know, when I did the research about the condition, just reading up about it and how it can, I mean, it's so complex.'

''I thought working with people with dementia was complex and hard, and again, you know, they are people that I think are really marginalised in society but reading about Huntington's Disease, you know, physically and the mental health aspect and with this lady that was the thing that actually really impacted on her mental health significantly. I think even the Huntington's adviser said to me that she was one of the more extreme cases really, with how it had taken a grip on her, and potentially, was destroying the family. She wouldn't talk to her mother because she'd had these thoughts and delusions, she wouldn't talk to her best friend but they never gave up on her either, you know and that was, again, it showed you that her character, cos she, it was clear that she was a really kind, caring and clearly a wonderful mother, you know, look how all of her children have turned out, just amazing really. Erm, yeah, so I'm happy to never give up on them."

Poppy Williams' approach was deeply valued by Marjorie and Timothy. For the first time in for many years, the children found a health professional who truly understood their situation. In understanding the disease, Poppy didn't lose sight of Jen Walters as a person.

Timothy emotionally recalled receiving a phone call from Poppy Williams: *'Look, Tim, I just wanted to speak to you on the phone, I just wanna let you know that I'm not gonna give up on your mum.'* Timothy recounted, *"Honestly I just burst into tears cos I was like, I felt, you know, at the very least this is someone that knows how to communicate, you know and knows how to, you know."*

While Jen Walters was confined in the psychiatric hospital, her mental health assessments indicated that she didn't have the capacity to make decisions about where she lived. Poppy Williams found herself in a dilemma, in that providing a home placement for Jen Walters, this essentially would make Marjorie Walters, who was still living with her mum, homeless.

Poppy felt she needed to protect Marjorie: *'What was even harder was she'd come to that point where she had disengaged and she wouldn't talk to anyone, the only person she'd talk to was her daughter. So then I had the battle of I understood why professionals wanted the daughter to communicate with her mum but I felt the need to protect the daughter because she was experiencing watching her mother go through this and then to have that responsibility placed on her, with all the other professionals I just didn't feel was right but I felt like I was kind of the only voice for them, if that makes sense?'*

'I always, I think what's been really surprising to me was I always felt I hadn't been able to do enough, you know, I worried I let the young daughter down, cos her mum going into care, technically made her homeless, it was just awful, it was just awful.'

Poppy had to weigh the situation very carefully. It was one of the most trying times in her career. She couldn't fully determine if Jen Walters would like to move up north close to where Timothy lives and where

a specialist HD placement had been identified. Poppy explained, *'So we just kind of had to weigh it up and the way I weighed it up was I looked at her support network and her strongest support network was her children and they were the people that she was still engaging with and so knowing where they were gonna be and I trusted that they were gonna be able to still give that support because there was no question about their love and their dedication for their mum, that that felt like that was the right decision.'*

'It was about I suppose taking that positive, it's a positive risk. but yeah, usually you'll get an independent mental capacity advocate who will come and meet with that service user, they will talk to them about their views and wishes and they will put that forward but they will also be overseeing to make sure that the decisions that we are making as representing the local authority, that they are in that person's best interests, that we have explored the least restrictive option and I felt like I couldn't even get those kind of safeguards for her because services just weren't working with her. It was very difficult.'

Another major obstacle came when the specialist placement, Driftwood Nursing Home, hesitated to accept Jen Walters. Citing her disengagement with care professionals and her case history of physical aggression, the home manager stated that they couldn't be sure that she was type of person they could appropriately support and work with. Poppy Williams pleaded to give Jen Walters a chance, and the nursing home eventually agreed to a trial placement. After the funding had been agreed, Jen Walters was moved out of the psychiatric ward. The Walters family relocated over 200 miles. Consequently, Marjorie went to a university that was close to her mum's nursing home and to where her brothers lived.

Within a few weeks of settling into Driftwood Nursing Home, Jen Walters gradually started talking. She slowly engaged with staff and started to eat again so that she was able to obtain the optimum weight. She accepted the support of the nurses and allied health professionals, such as the dietician and speech and language therapist, to address her needs. She also engaged with a physiotherapy programme. Gradually, Jen regained her physical health.

Jen's children visited her regularly and after a couple of months, they took her out into the community. For Poppy Williams, Jen Walters' case, which she handled over seven years ago, will stick with her all throughout her professional life: '*They got a little bit of their mum back and for me that was amazing.'*

As Timothy recounted his mum's HD journey, he was indebted to the efforts of Poppy Williams in finding the suitable home placement. But what moved me in our conversations was how he appreciated the way Poppy Williams communicated with him. *"Just having someone that, that gives, you know, someone there that says 'look, I get it, I get it' and even just someone that actually looks you in the eye and says 'I understand, go and wipe your nose' and it can, honestly it does, erm, yeah, it does make the world of difference."*

Similarly, Marjorie described Poppy Williams as *'probably one of the only health professionals who really saw my mum. I don't know where we would be without Poppy Williams to be honest because I genuinely think she was, you know, a bit of an angel in that respect.'*

What is important for Marjorie is that she finally had her mum back: *'I kind of sometimes forget that she is, that she has Huntington's because she's very, her delusions have obviously gone and she's very present, which is, you know, the biggest thing, that's the best thing you can ask for really, there, and she's so present and she asks me questions, she, you know, just spending time with her is, is, for me, I find it incredibly nourishing and, I don't know but I think she has changed a lot from how she was when I was living with her but I'm just, I'm so grateful that she has this kind of health care around and obviously Poppy is someone to thank as well as people at Driftwood Place Nursing Home, of course.'*

Marjorie said, 'We can actually have time together where it's much more meaningful and she's there, present, with me. Which is all I can ask for really.'

PART THREE: RESIGNATION, I AM HAPPY HERE

'I am happy here,' Jen reassured me. In our extensive interactions, Jen and I would often pick up our conversations with her personal mementos in her bedroom. At the corner of her bedside table is a stack of photographs and postcards. I flicked through the captured smiles and letters, then glanced at Jen finding her smiling back at me. She ardently spoke about her recent activities and her children visiting the nursing home.

The key aspect for Timothy and Marjorie is the right environment and people to provide care for their mum. For once, Marjorie explained, 'She's *been so comfortable and so herself, so happy and it just, it goes to show how being in the right environment is just so important, having the right, having people around you who understand you is so important for someone with Huntington's to live a life, well to live a life of quality. It's a blessing to know that she is looked after now. I think she's more herself than ever really.'*

Jen has regained her previous interests of painting and drawing. Timothy recently shared with me photographs on his phone capturing Jen and him drawing together. In an A1 sketch paper, they drew pastels of a vase with spring flowers. A clumsy mosaic which imbues colours, of amber bulbs and wilting khaki leaves. Jen shared many drawings in her room. As she unveiled and shared her life, she spoke richly about these artworks and photos, *'I've got loads of happy memories',* she exclaimed. One by one, she told me what each artwork in her room reminded her of. These are the memoirs that Jen and her children chose to keep.

Amongst these, she referred to a black and white photo of Timothy and another woman. Jen told me it was taken on a trip to Brussels to visit her cousin. She liked that picture because *'I've got everyone laughing'.* The picture was taken on a sunny day behind a canal with cafés littered with baskets of petunias. Then, Jen pointed to an imposing poster on her bedroom wall, a crude black silhouette of a man. He lies in a deep blue blanket scattered with yellow sunbursts like explosive dandelions. Between his chest was a distinguishable red dot, it resembles like a heart piercing through a bleeding wound. Jen said she picked it up in a museum shop. She told me that Henri Matisse is her favourite artist, and she attributed her

enthusiasm to the shapes and bright colours of his paintings. I later learned that the man in the Matisse print was Icarus, who flew too close to the sun on wax wings and plummeted back into the sea.

Jen showed to me a striking resemblance of Matisse with her own artwork. Set in a 4X4 canvas, in a backdrop of vivid colours is a black silhouette of an animal paw. On the lower right side of the picture, the name 'Charlie' was written. Jen told me it was her cat. She had made the watercolour art in an activity session in a previous care setting. She said she couldn't remember the name of the facility or the date it happened.

Jen also pointed to a white frame on her wall. It was a print of a young boy chasing a glaring orange, reminiscent of a ball half the size of his body. On closer inspection it was apparent that the boy was dressed in a red football kit. The artwork was made by Timothy, who attributed his creative talents to his mother. For Jen, the picture reminded her of her youngest son, Francis, playing ball. Jen said, *'It brings back very good memories of the kids.'*

But her favourite was the arresting painting in the middle of her room. Jen said it was painted by her friend in London, *'I have to pay her for this',* she laughed, *'I love it'.* It was a silhouette of a woman in a yellow dress. With one hand the figure is carrying a child in a white loincloth, while in the other hand she holds an oversized blue umbrella. The woman is wading through the rain, sheltering the child from the dark skies.

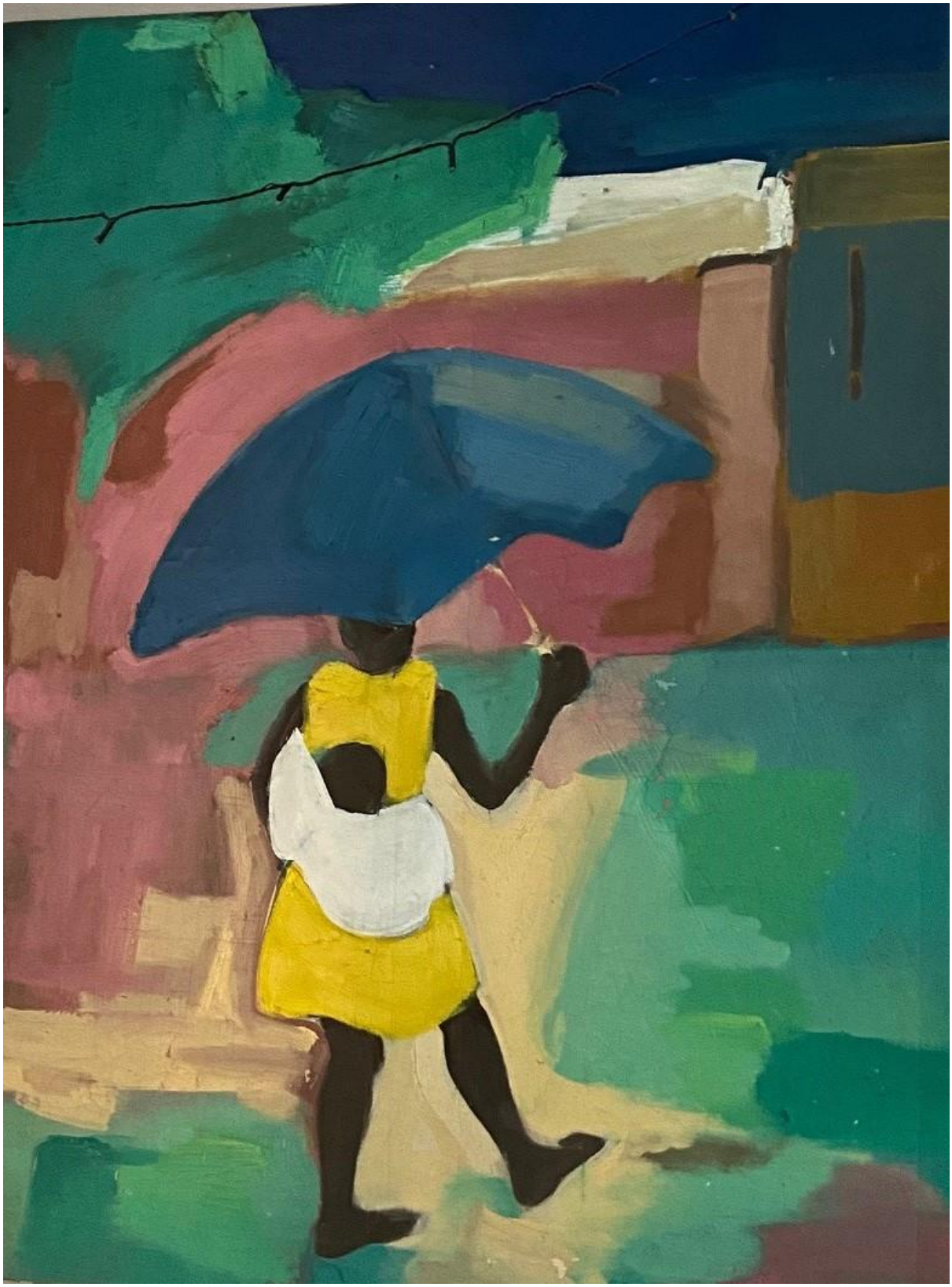


Figure 23. Jen's favourite painting

EPILOGUE

It dawned on me in my many interactions with Jen that she is keener to talk about different aspects of her life than her disease. Perhaps this was all that Jen was trying to reveal. I finally realised that when telling her life story without specific prompts she eloquently evoked it through arts, crafts, and personal keepsakes. For Jen these vestiges preserved her memories, which refutes her initial assertation of *'I couldn't remember much'.*

In my last research interaction with Jen Walters in a video call, I knew that the next time I would see her in person, her HD would have inevitably progressed. But a smile never left her face, she is happy where she is. She wilfully resigned to her situation. As Jen Walters could no longer do things by herself, she stolidly reconciled her fate and her disease to the care of her nurses and carers. By living in Driftwood Nursing Home, she has eased the worries of her children because her physical health needs are looked after.

For Timothy, Francis, and Marjorie they can create meaningful and lasting memories with their mum. Timothy informed me that when the Covid-19 restrictions are lifted, together with his siblings, he plans to take his mum in a hot air balloon trip. I am reminded again of the story of Icarus. But this time, the story isn't a solo flight, for Jen will be together with her children. As she faces the late stages of the HD storm, like the woman in her favourite painting, all she ever wanted was to protect her children and keep them close in her arms.

MARTIN TAYLOR

THE MEETING

It was a rainy Wednesday evening and the clock turned past ten. From the nursing home window came the sound of the heavy January downpour. I was hurriedly cramming the paperwork into my backpack when I felt a gentle tap on my left shoulder. I turned around to see a rotund man in his late 60's, standing at about 5'10" with brown doe eyes and seamlessly combed quiff. He was dressed in a blue Gingham shirt tucked into black chinos. He introduced himself as Martin Taylor, and quietly he said, *'I am a nurse, I have Huntington's.'*

It was the night when I had spoken about my research recruitment to the HD support group. The meeting had been held in a nursing home. That was the second time Martin Taylor had attended the support group with his wife, Pamela Taylor. The group leader had given me a space to talk about the PhD research. There were 12 people, some of whom had familiar faces having previously been my HD patients. Initially, I was introduced to the group as a physiotherapist, but I then explained that I was interested in wide-ranging HD experiences rather than solely on exercise therapies. When no further questions were raised, I left posters about the study.

Martin Taylor was holding one of those posters in his right hand. He told me that I must speak to his wife, *'I have trouble remembering dates,'* he added. When we started walking, I noticed his fitful movements. Martin's limbs faltered and his legs couldn't keep still. We approached Pamela, a blue eyed, coiffed blonde full-figured woman sitting with a cup of tea. We spoke about the research, and I encouraged them to read the information sheets. Pamela reached for her handbag and penned a number on the back of a torn shopping receipt. I said, *'I will call in a week's time.'*

RESEARCH INTERACTION

I called their home number after a week and the couple agreed to be interviewed a couple of weeks later. They invited me to visit their family home. Martin Taylor mentioned that afternoons were preferable as he was *'not too good in the mornings.'* I jotted in my field notes reflections on our first interaction (Fig. 1)

19th February 2020, 13:20, Bus 780 to Case II, MT

This bus ride will take me an hour and a half to reach MT's home. There's this ambivalence, the apprehension and excitement as this will be my first time to conduct research outside a familiar setting. My experience of interviews and focus groups were held in public venues, clinical or controlled environments. This time, it will be in a private home. I don't know what to expect when I get there. I suppose the narrative approach to research offers this more personal interaction. The control of space and time; the power is shifted to the participants as they take ownership of the interaction, the conditions are on their terms. I will be guided to where they want to be interviewed, at their comfort and convenience.

After all, I am interested to hear their story, what better opportunity to witness this is at the heart of where they live, where lived experience happens as it is. I want to encounter participants as they truly are, unperturbed by socially imposed rules and boundaries of a sanitised research environments. I've prepared everything I needed (fingers crossed!), I bought three pens in case the other two won't work (what are the odds?). For whatever the outcomes, well for one, this will be a new research experience.

Figure 24 Pre-interview notes

In the span of 18 months, Martin Taylor participated in a total of five research interactions. These were joint interviews with his wife, Pamela Taylor. There were occasions within the interactions when separate interviews occurred, Pamela would say *'I'll leave you to talk with Martin'*, or in Martin's words, *'I'll put you through to Pamela, as she remembers more than I do.'* But all interviews started and ended together. The research interaction was divided into two phases: before the Covid-19 lockdown and after. The first interview was held in the family home and the second interview was conducted after the HD support group meeting. The planned observations of their health appointments and social activities did not occur due to the pandemic restrictions. The next two interviews took place remotely, and the exit interviews were, once more face-to-face.

MARTIN TAYLOR AND WIFE, PAMELA TAYLOR

Martin Taylor was born into an Irish immigrant family in the industrial north of England. Both his parents worked in factories. Their only child, Martin, believed, *'family is everything'*. Inherently, he took the responsibility over their family affairs. As Pamela affirmed, *''Martin took their finances in hand when he was 14, paid the rent, paid all the bills. His dad just went along with the flow."*

Martin started working in the civil service at age 17. Aside from his commitment to work, he was a devout supporter of his football team, the love of sport he shared with his dad. Raised by religious parents, he also attended church activities and went on pilgrimages. At 33, he met Pamela on a spring trip to Lourdes, France. She was with another church group. He was drawn to her *'confidence'* and she was besotted by his unperturbed rocker type appeal. While she described him in *'leather and bikes'*, and she was the type to dress to the nines, they *'clicked'. 'We are very opposite, in our views, personalities and everything',* said Pamela, '*But we bounced off each other'* and they '*just got on'.*

Pamela Taylor, the same age as Martin, was born in Michigan, United States. Her American mother met a '*lovely'* man and they emigrated to England when she was three years of age. Six months into their relationship, *'I asked Martin to marry me',* Pamela laughed. She told him, *'Listen Martin, if you want to be friends, fair enough, but I want to know is it more serious?'* That pressed Martin to ask for Pamela's hand and they got engaged by Christmas. As soon as they found a house to move into, they set the marriage date. Pamela explained, *'Because there is no such thing as living together with our families, not a chance. We found the house and got married six weeks after.'*

Martin and Pamela described a *'happy married life'. 'We always do things together,'* Martin repeated. They went on yearly cruise trips abroad and continued with their religious activities. In the first year of their marriage, they set up a charity for disabled people to go on pilgrimages or holidays. After they observed that their churches *'did not specialise in disabled needs'*, also they found that non-Catholics couldn't join the organised church trips. Although '*demanding'* of their time, the couple found fulfilment in fund raising activities and they *'were almost out every weekend'*. Eventually work, and other personal circumstances, got in the way and the charity ended after nine years.

Soon Martin returned his attention to his aging parents. Martin had '*no problem with the old man',* but Mrs Moira Taylor was known to be imperious and feisty. The younger Moira *'was very good looking and attracted unwanted attention'*. Whilst working in the factory, *'She was on the line with all the men. She was a strong woman.'* Martin described how his, '*Mum used to enjoy going dancing. Mum and dad used to go on [ballroom] dancing. But dad, he went for the easy life. If you upset mum, then you had to pay the price of how she was going to respond. So, he tended to disappear to the football game. If mum kicked off, they used to drag me in because the doctors had turned round and said the only way you can get my mum to respond is to have me within earshot or be in the same room. It was, I found her alright, but she was hard work.'*

When Martin's dad passed away, this led his mum to live in a nursing home. Moira's mobility and memory deteriorated, coupled with '*challenging behaviour*'. The move to a nursing home was a decision made by Pamela and which *'wasn't up for negotiation'. 'We wouldn't have had a marriage and it was just a No!... So, she went into a home or [Martin] went and lived with her [laugh].'* Martin admitted that his mum was '*difficult*', cantankerous, and aggressive, and that *"You had to pin my mum down to control her, so it was, you weren't very popular when it came to any relatives or any friends if mum was coming along. You were expecting trouble."* Pamela agreed that Moira Taylor *'didn't disappoint'.*

Pamela said*,* '*We got through our wedding day without her, without an incident. She phoned me up at 3 o'clock in the morning before I got married calling me for everything. She didn't want me to marry him, she didn't want him to get married at all but on the wedding day he warned her. She was scared of Martin, and she took notice of Martin.'*

Moira Taylor spent the rest of her life in institutionalised care. *'I feel guilty as I have been looking after my mum all my life',* Martin reminded me. He witnessed how his mum's mental health subdued her and had impacted family relationships. Martin's experience of caring for his mum enthused a late career change to nursing. He went back to university and qualified as a mental health nurse before he reached 50 years old.

THE ROLES WE MAKE AND THE TABLETS WE TAKE

PART ONE: CHAOS

HIS BREAKDOWN

'I was hoping that a bus or a car would hit me'. Martin had a mental breakdown at work. At the time he was working with the community mental health team: '*Anxiety turned into depression, and I was saying to one of the occupational health; 'it's a shame that you can't turn round and take me or anybody else out of this'.* Ironically, he revealed, *'I was nursing and caring for people within the community, and I became unwell. I wanted to kill myself at work.'* Martin spoke with vulnerability: *'I've got anxiety, depression, depression no, no, no, you don't like that, it's' horrible. The anxiety you will build up your discussions with the teams, and hope that it doesn't go to depression but on occasions it did.* Martin paused, and said, *'I became, suicidal with my thoughts and actions and people. I was quite a sick person*.'

The first breakdown happened about ten years ago, the beginning of Martin's depressive episodes. While Martin believed his depression was a result of a build-up of personal life experiences, Pamela disagreed: *'He started to do extra work for the fellas in work and everything started going wrong. He started doing all the double shifts and that's when everything started going wrong.'* Pamela recounted this coincidence with the sudden change of his behaviour. Pamela used to own a café business, in which, she asserted Martin was *'the brains behind everything'.* She recalled around the same time, '*I was going through, we were going through selling the business and I never had the support I'd had all the years previous, he left everything more or less to me, it wasn't Martin's style. All of a sudden it was left to me 'whatever you decide, whatever you decide'. The day that I sold the business, and I was going out with the girls from work, we were taking them all out for a big night cos we were closing, and I'd come home from work that day and I just remember everything was wrong. Everything was wrong.'*

Pamela detailed Martin's many peculiar behaviours, amongst them when he surprised her with a phone call that informed her that he had paid a deposit for a *'chaperoned'* business class luxury cruise. *'That's the first time he's ever picked a holiday without me and it was, like, over the top holiday and by the time I come from work to home, he wanted to cancel it, he didn't wanna go.'* Martin's reason was: *'They said that they would transport you from your house to the ship and what happened is I couldn't get out of my mind the thought of sharing a luxury coach with other people.'* As Pamela explained*, 'So he'd taken his time on what he'd done and then a couple of hours later he brought me right back down again.'* Martin lost £1,600 of non-refundable deposit, *'And then cos it was with Virgin, he decided to take a dislike to Richard Branson*

cos he couldn't get his money back and I got it for six months every day, all day about Richard Branson. Called him all the names, you wouldn't believe the names that he came out with.' Distraught by his actions, Pamela issued a stern warning and '*He swore that he wouldn't ever do anything like that without me and I've told him if he does, I'm gonna take total control of the money.'*

This was just the beginning of Martin's indifference to Pamela, and on many occasions, he would verbally assault her for no apparent reason. The frequent rows escalated to talks of the couple separating and Martin got his own flat. *'We've had a really good married life. We have, until Martin took bad, and he was just getting so unpredictable, so argumentative, so insulting and the language sometimes that he called me'.* Martin acknowledged this in our conversations: '*I behaved terrible Paul. I was very ill, I was getting very bad, suicidal.'* Consequently, Martin came to the attention of the community mental health team and was referred to a psychiatrist.

HER BREAKDOWN

'I pulled up, I was driving somewhere, and I pulled up and I just cried my eyes out cos I couldn't cope with the way he was carrying on. He gets obsessive thoughts, and I can't even remember, it might have been, it was just the straw that broke the… [long pause] and I phoned up his consultant and the hospital, and there was no-one there to help me… there was no-one there at all'.

Pamela continued, *'I was crying and crying and going 'I can't cope with you', you know, and I phoned my doctor's that night and he said to me, 'you need to have a word with the consultant' and so the next time we went I sat there and to be honest they were useless. He was useless, all that Martin's consultant said was 'how have you been doing? When do you want your next appointment?' and I sat there, and I went "Is this it? Is this what I've got for the rest of my life?" you know, ''you tell me to go to day centres and this, that's not the answer".*

Martin took the view that half a decade *'of taking tablets and scheduled health visits made no difference'.* Martin's psychiatrist gave him *'leaflets on men's health groups'* and incessantly advised him to *'go to the gym',* but Pamela observed her husband was becoming very hostile. *'He was a raging bull. He was horrible, just horrible. Our marriage was breaking up cos he could be that horrible. Our marriage is going because he's not happy with me, I wasn't happy with him. We couldn't agree on nothing; '* Pamela repeatedly reached out to the Crisis Team when Martin was having nervous breakdowns, to which they replied, *'There's nothing we can do'.*

Martin's enmity and depression were becoming worse, and Pamela reported she constantly received threats from Martin saying, *'I'm gonna kill myself'* or *'I'm gonna leave you'.* This difficult period Pamela described as *'six months that we were in each other's throat'.* Pamela described one day thinking *'I am not having this! This is not my life'.* Pamela reported her own strong feelings, expressed in the words, *'I love him enough to wanna do something about it'.* She took matters into her own hands and *'dragged'* Martin to her own family doctor. Her doctor who saw Martin for the first time, *'He said right away, your*

movement was not right'. Martin was accordingly referred to a neurologist who specialised in movement disorders.

HUNTINGTONS: THE NAME THAT SAVED OUR MARRIAGE

'We walked out of the clinic, and we were both in, like, a little bit of a daze'. Pamela recalled that moment as she vividly described that day. The neurologist, Dr Donato, had clinically diagnosed Martin with Huntington's Disease (HD). '*He reckons he had HD about ten years ago.'* This was about the same time when Martin had his initial crisis. *'Now we don't know whether the breakdown come first or the Huntington's come first, one kicked off the other, we don't know.'*

'Well at least I know why' Pamela continued*, 'I used to say to him, I love you, but I don't like you.' 'Well, we've got a name now, we've got a name, and that was my, Martin was not just being horrible.'* She looked at Martin and said, *'You're not being awkward, you're not being, you know, we've got a name for it and to that to me was…'* Paradoxically, Martin's incurable diagnosis could be said to have provided something like a relief that she thought, '*the biggest difference was having a name. Knowing that he wasn't, I really thought we were going to have to break up.'*

'It all started to make sense'. Pamela revealed to me that, *'Things tend to fall in place, and do you know what Paul, I, I think Martin having Huntington's saved our marriage…I know it's not Martin, it's the Huntington's. I'm more accepting.*

Pamela described the outlook she adopted in the following terms*, 'Let's do something about it and let's find out as much as we can. I went on the internet, which was very frightening but at Martin's stage, I found out as much as I can about it. We're just gonna take every day as it comes'.* Dr. Donato prescribed *'new tablets'* and after three weeks Martin noticed it '*appeared to take a lot of anger away, it also stopped the movements.*' In Pamela's observation*, 'he was like a different man, his temperament changed and everything.'*

Martin recalled receiving his diagnosis: *'Dr. Donato turned around after seeing my erratic movements that I have to put up with or what I will be looking forward with Huntington's. I can see myself getting worse and I get worse, I can manage what I'll become. My only problem is, I have cardio-vascular illness that might trigger my Huntington's. Hopefully, if I remain the way I am I'm delighted with that.'* Pamela further added, *'I think I've figured out now with the tablets he's on, he's better now.'*

PART TWO: QUEST

THE BEGINNING AND END OF THE LINE

'You can quote this for nothing, I was like a Huntington's virgin. Because apart from assuming that mum had Huntington's, I'd no involvement at all.' Martin described coming to terms with his condition. *'I was*

listening to people who told me because I'd never, even in the medical profession I hadn't come across anybody. It was not too known about. Until we joined the support group, I didn't realise how many people have Huntington's.'

Martin had been unaware that his mum had Huntington's until recently, when they looked up her death certificate. *'The first we heard about it really.'* Equally confounded, Pamela said, *'Where did this come from? Cos in the old days, she went to one doctor, and she used to get shock treatments. She was that violent that they used to take her in for shock treatments. She had mental illness, end of story, you know… She was not on them tablets',* Pamela added.

Pamela explained that Martin *'started with movements exactly like her'.* Moira Taylor *'couldn't stand in one place, she always lifts her leg up, like her heel up her bum'. According to Pamela, this was 'the kind of jerkiness'* that Martin replicated. *'He permanently sat like that, that the couch worn out; it was bad. I used to say, 'Martin, stop it, you're like your mother.'* Pamela added, *'to be honest, I think I got used to the movements'.*

But it wasn't the spasmodic movements that Martin remembered. *'She was quite violent mum'. You had to pin my mum down to control her… ECT (electroconvulsive therapy) is the only option if I become unwell or deteriorate'.* Martin turned his gaze to Pamela and said, '*It's the only way but Pamela is not very keen'.* Pamela asked me *'What do you think of shock treatments, Paul?'*

I kept my opinion as Pamela interjected, *'I don't believe in it. The tablets are different now when your mum was still alive, and the doctor was about 102 that used to look after her, you know.'*

As I inquired into the presence of HD within Martin's family, Pamela responded, '*The line stops here [laugh]. I got pregnant when I was 39 and ehm, it turned out the baby was in the fallopian tubes, and I had a hysterectomy. I was upset at the time. I would have liked kids probably but because I couldn't have them it was just one of life's things'.* Pamela turned to Martin and said, *'We knew there was something wrong with your mum, we never knew what it was, but we knew there was something there'.* She continued*, 'neither one of us was that bothered on children. So, when I got pregnant and ended up losing the baby, it wasn't a big deal. That sounds cold doesn't it, but it wasn't a big deal'.* Pamela distinctly remembered her conversation with Martin's neurologist*, ' When we went to Dr Donato, he said, "have you got any children?" and we said 'no', he went "good", and he went. You know when someone wants to take something back but it's obviously how he felt'.*

Pamela reasoned that *'things were meant to be',* she continued that, *'We've got two businesses and I wouldn't have been able to do that if I would have children. So, your life's mapped out for you and I'm a firm, firm believer that Our Lady looks after us and Martin is the same. I know we go on a little bit about religion, we're not God preachers or anything like that, but we're both firm believers Our Lady will show us the path.'*

The interview I have been describing constituted the first time that Pamela and Martin had talked about HD as a couple since Martin was officially diagnosed over two years ago, and he admitted, *'I am learning about it, and I don't know which way I am going to go Huntington's wise.'* Martin compared himself to other members of the HD support group, and said, *'I think I'm a little bit surprised that I am going to go a lot worse than what I am'.* However, Pamela disagreed that it wasn't helpful for Martin to compare himself to others, *'I don't like seeing advanced patients. Nothing personal but I don't like to see what might be'.* She reassured Martin that, '*You're actually gone better in the last 12 months. But I think you are getting better. Now, whether we were getting more used to it or not but no, I think you are getting better'.*

Although the couple had started to get involved with the Huntington's support group, Pamela still insisted *'I don't mind where we go but I don't wanna be, I don't want to live my life with Huntington's, I wanna, I can't find the words...'* and Martin interjected, '*Wash your hands of it?'.* Pamela answered, '*Obviously, we're not gonna wash our hands, you've got Huntington's'*, but she then continued, *'I don't want it to be my only thought, I don't wanna live with Huntington's. Does that make sense?*'

While Martin likes being in contact with the group so he could find out *'what I will be able to cope with and where I'll need some help'*, Pamela asserted, *'we also have a life'* and *'leave it will until it happens…wake up in the morning and see how you are in the day'.* Pamela reasoned, *'I personally think that with Martin's age now, we're just gonna live like two old fuddy duddies. We're gonna live our life out, you know. But I know in my physical state I can't physically lift Martin, I can't physically bath him, physically feed him and everything like that. Physically wise it's wait and see.'*

SOURCES OF SUPPORT

Currently, Martin is under the care of his GP and of the Neurology Consultant. Whilst they get no *'official support*' from health professionals or external agencies with Martin's care needs, Pamela's siblings offer them valuable assistance. '*My brother has started coming up once a week regular now, I know I can phone him up and say' I need you and he'd be up'. My sister's here to talk to.'* Pamela mentioned that '*If I needed anyone and wanted answers'* in relation to HD, that her '*first call'* would be the HDA advisor, they would also phone their GP when needed. Martin responded, *'We try not to ask anybody, we try to do it ourselves.'* He asserted that *'We've got our faith. We've been linking church with the way forward'.* Following Martin's diagnosis, the couple reignited their devotion to church, not only did this fulfil their spiritual needs but also provided social support. More so in the Covid-19 lockdowns, where they continued going to mass through live streams and '*have a little gab with them after'.*

THE BADGES I WEAR

'I don't know whether I prefer the Huntington's badge, or the mental health, anxiety, depression badge. I see there's like a common badge between these illnesses, Er, I don't, I don't feel as though I have a mental illness, but I know I have. So, I've got to try and think about that.'

During this interview Martin reassured me that, *'At the moment I can manage and cope quite well,'* however, in all our interactions he consistently voiced concerns over his precarious memory, '*I know it's dangerous but when you can't remember something, and you get impatient and frustrated because you can't do what you're thinking about what you're going to'.* Martin said, *'I've been trying to join the memory clinic but, er, it didn't happen because people will just say, 'Oh you've got Huntington's'*. He asked*, 'Am I expecting too much Paul?'* in a somewhat defeated tone.

'I've been discharged from the community mental health. I know that I still have erm, depression and anxiety traits and I feel annoyed they discharged me. They should have a meeting with me at least, but they don't bother. They don't talk to you; they don't talk to the likes of me'.

Martin's psychiatrist had also discharged him, that according to Martin, his psychiatrist remarked *"Now you're with Huntington's you don't need to see us".* According to Pamela, the neurologist questioned this, by asking, "*Why was it stopped, I think you still need to see a psychiatrist*". The apparently conflicting messages of consultants dealing with different aspects of his physical, psychiatric, and cognitive HD symptoms led Martin to comment on his own self-reliance: '*We've always looked after ourselves, haven't we? '*

Pamela answered, '*To be honest Paul, I'll go anywhere if I think it's gonna help Martin. We'll try every avenue. I will try and have anyone in the house'.* Pamela later revealed her own health issues, such as the excruciating pain from '*bad arthritis'* in her hands and back, for which she *'gets injection every six months'.* Her unwillingness to fully agree with Martin probably comes with the recognition of her own failing health, that perhaps she was more willing to accept support from other people.

PART THREE: RESIGNATION, WHAT I (WE) HAVE GOT

For Pamela, *'I think the biggest thing at the moment is accepting. Acceptance'.* Which Martin interrupted, *'Of what I've got'.* Then she corrected him saying, '*What we've got… and live with it'.*

In our conversations, Pamela confided thoughts she would otherwise have kept to herself. *'There's one thing that might help with your research Paul, and I wouldn't admit this to many people. From I'd say that month of his mental breakdown, we've never had any sexual relations at all. It doesn't bother either one of us. Whether it helps your research or not, whether you need it or not. I don't know. But if you're gonna get the picture, you may as well get the full picture'.*

Pamela reported the vagaries in their marital relationship: *'Martin had terrible movements and it kept me awake. We slept together, we were married, and you know we slept in the same bed. One day in his sleep, he punched out and gave me a black eye. It was in his sleep, he didn't, you know. So that's when we decided we were gonna sleep in separate rooms then'.* Despite the changes, Pamela maintained that *'it's no big deal, no. It wasn't a big deal'.* All through their marriage, Pamela has always done things for Martin: *'He doesn't do a job round the house; this isn't a complaint it just a fact that he doesn't even wash a cup*

after him. Martin's never done housework, never cooked, because I've never let him. That's probably a little bit my fault because I've always done it.'

Martin's loss of functional independence had much impact on their home life, but Pamela confessed *'What frightens me more than anything is me dying and leaving him. It scares the shit out of me because he wouldn't be able to manage his tablets, where to get them, that does scare me. I haven't quite figured out what to do yet, but I will do. I need to sort all his medications, where we get it from and everything.'*

'I don't know, I don't let him do, right or wrong. I've taken too much off him, do you know what I mean? But it's what I've done all my life' Pamela said in an exasperated way, *'what's Martin gonna be like in ten years? Am I gonna look after him more? We don't know, we just don't know, and that's why it's each day as it comes'.* Notwithstanding her impending concerns, Pamela consoled that, *'We've adjusted. Life isn't bad Paul, all doom and gloom. Life isn't bad, I just miss my old Martin.'*

'If I continue the same it shouldn't be a problem', Martin suggested. However, he understood the inescapable worsening of his HD symptoms, *if that stage presents itself then I can see me going to the nursing home, managing around there, I'd also be able to see the Huntington group once a month, so I'd still have that link. But at the moment my heart is good, and I prefer not to think about it because I don't want to upset the routine we have at the moment.'*

Martin conceded, *'I get myself annoyed because I struggle remembering people's names and faces and I can't remember my telephone number and I don't want it causing me embarrassment,'* but Pamela consoled him that his memory could be due to *'old age'*. Martin also disclosed to me that he is *'quite bothered about Pamela's legs and back, her rheumatoid arthritis.*' However, despite his worries, he asserts, *'I feel mentally good. I've not got any structure in my life, but I feel great at the moment Paul, So I don't push it'.*

'I have very little anxiety, er.. and my biggest problem is that I want to do, do more but I shouldn't be. I should be happy with what I'm doing at the moment and the interactions that I've got. At the moment, I'm very, very stable but when depression appears it's not, erm a good sign'.

EPILOGUE

I called Martin and Pamela in the Spring of 2021. The UK was still under some lockdown restrictions, which required that people should continue to keep their *'social distance'* from anyone who is not in their household or support bubble.

When I heard their voices, I felt their elation; *'We are seeing family again; our nieces were coming up and we're starting to mind them again'.* Pamela referred to the Covid-19 vaccinations, *'we had both our injections, and we are ready to rock and roll [both laugh] whenever rock and roll is gonna come out'.* She recounted, *'We have been very lucky that our church has a big garden',* and the couple were able to attend

the sporadic outdoor church services. Whilst the support of their families and friends from church continued, their health appointments on the other hand, had been put on hold. For over a year Pamela had been waiting for her *'spinal injections',* and Martin hasn't heard anything from the mental health team, nor does he know the date of his next HD review. Their health consultations, which in Pamela's words '*We do desperately need,'* had not occurred.

Additionally, Martin's nightmares had returned. His sleep had been disrupted and his moods had *'been up and down, but when he is down, he is really down'*. He was having *'really bad dreams about dying, death and other people'.* When these *'down days'* occur, Pamela said this makes him '*very tired for the rest of the day,'* that *'he overthinks everything'* and *'needs picking up'.*

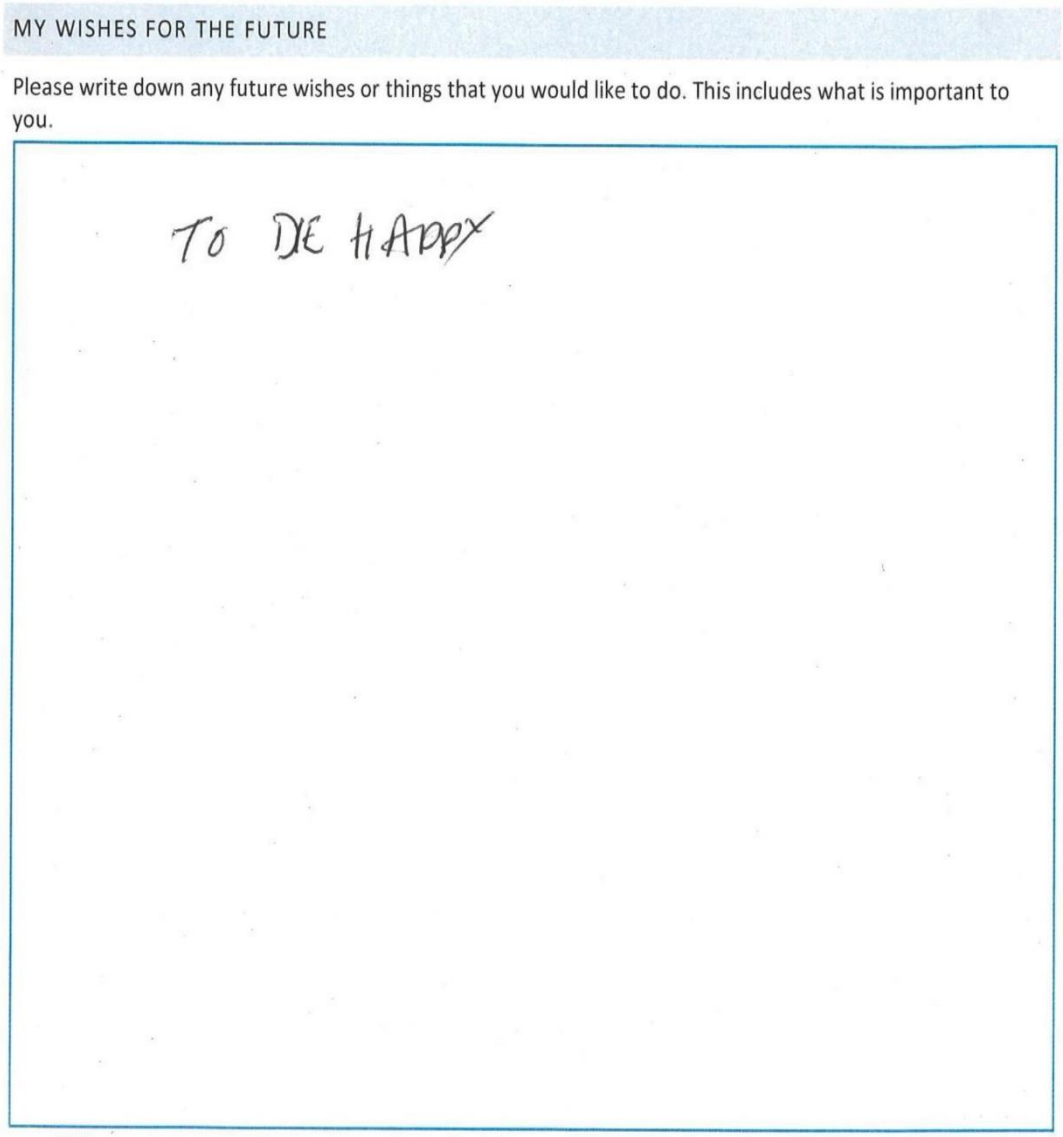
Other than the sleepless nights, Martin deemed there wasn't much that had changed since I last saw him in person*, 'I feel quite healthy Paul*'. Pamela disagreed, pointing out that *'he has put on too much weight'* and *'he is more unsteady with his feet'.* Martin's unawareness of his movements and evident lack of coordination meant Pamela had to assist him with changing his clothes, also with *'helping him shave, have a bath, help in the shower*'. Pamela reasoned, *'to be honest, these come up slowly that basically we got on with it'.* Martin had also given up his driving license, and he had revoked his club membership and season ticket, and it was reported that *'it upsets him too much when he thinks about it'. 'It is safer Paul*', Martin told me, reflecting on the fact that he could no longer physically drive nor brave through the raucous crammed football games.

Pamela described to me in our later conversations the full extent of her caring duties. Rather than feeling burdened she facetiously remarked to her husband, *'You're a nuisance but sooner have you with me'.* True to her words, I was reminded by what she confided in our initial interview *'If I can cope with Martin at home, no way is he going into a [nursing] home because he's got Huntington's'.* However, Pamela acknowledged that *'If I can't, get to the stage where I can't cope with the illness and looking after him, we can bring people in. If that gets too much, well we've gotta look for a home but I'm hoping that never happens'.*

Martin characterised his relationship to his wife as: *'I need her'.* Pamela in return looked at Martin in the eyes and said, *'You know, we need each other'.* Pamela referred to their future living with HD, saying '*Nothing's written out for it, it's gonna be, it is what it is'.* Pamela revealed that she had Martin's *'power of attorney' so* that when he *'lose his capacity'* she can *'speak in his behalf'.* Furthermore, they had both planned for their impending future with '*their will is in a will safe'*, and *'cos we got no children we've arranged our funeral, how much it's gonna be and we've paid for it'.* In what seemed to be an attempt to justify her actions, Pamela asked me, *'I don't know what you think Paul, have we done everything right up to now or is there anything I need to do?*'

There was a sense of resignation from Martin when he nonchalantly replied, *'I know its gonna happen eventually*'. I couldn't exactly pinpoint what Martin was referring to, whether it was losing his memory, his

declining physical abilities, the onset of his depression, his thoughts about living in a nursing home or his recent recurring nightmares on death and dying. I returned to the transcripts and other documents hoping to find an answer, only to be reminded of Martin's resignation, for which he wrote…



MY WISHES FOR THE FUTURE

Please write down any future wishes or things that you would like to do. This includes what is important to you.

TO DIE HAPPY

DANIEL CARRAGHER

THE EMAIL

His email arrived the day before our scheduled interview. *'We lost mum last Friday. I'm really sorry to do this at such short notice. Can we postpone the interview in the next couple of days?*' Daniel Carragher added, '*As you can see things are still up in the air. I am sorry again.'*

My initial thoughts were of disbelief. A few weeks previously Daniel had written that despite Covid-19, *'We are all still thankfully well here. Mum, dad, and I had been put on the shielding list for 12 weeks '.* My second thoughts were: *'I should be the one saying sorry, not you. I am sorry to hear about your loss. Dealing with your illness, your mum's death, and the risk of Covid-19'.* I reflected on when the ideal time might be to conduct the interview. Would it be right to talk about his experience of living with HD, the same disease he shared with his mother? Would it be too much for him to deal with grief at the time of the pandemic? Would talking about his illness cause further distress? These questions preoccupied me while I thought through my reply.

I responded to Daniel to take his time, and that I completely understood if he chose to withdraw his participation. However, Daniel reassured me*, 'The funeral is next Thursday, and it should be easier after that. I still want to do this, so the week after is ok'.* I gave my condolences and directed him to support services, should he need them. I reminded him that I could be flexible with the date, and that he was free to message me at any time. Two weeks later I received another email from Daniel saying, *'The funeral went well as I suppose these things can. There were loads of people outside when we left, and then outside the crematorium which said everything. The funeral itself was strange due to only ten people being allowed in'.* He had ended the message, *'I'd like to speak to you tomorrow if that is okay. I look forward to it'.*

RESEARCH INTERACTION

Daniel Carragher is a Caucasian man in his early 40's. He towers at 6 feet and is heavily built with blue eyes and auburn hair. Born in the northwest of England, he speaks with a distinctive regional accent. He has always lived in his hometown, and he comes across as a placid and easy-going man.

Daniel heard about the PhD study from an HDA advisor. He expressed his interest and we corresponded through email. It was the beginning of the year 2020 when we first exchanged messages. I sent him a link to the study website (Appendix F) and consent forms were sent through the post. We initially agreed to meet in person. However, a week after our planned meeting the UK Prime Minister announced that country would be placed on a national Covid-19 lockdown.

Daniel participated in the requisite of four interviews over the 18 months of the study. The first three interviews were conducted remotely due to the Covid-19 restrictions. The penultimate and exit interview

took place in person. This was after the local restrictions had been lifted and the study was granted its third ethical approval to resume with face-to-face research activities (Appendix M).

On that day, Daniel was waiting for me outside a shop in a busy high street. He said that he had been early and had walked around town. He was wearing a black sports jacket, blue jeans, and tired white trainers. He was carrying an Asda plastic bag filled with documents that he wanted to show to me. At that time, the Covid restrictions only allowed people in separate households to converge in public areas. From a distance, I noticed his choreatic stance. He had assumed the characteristic HD sway with which I was all too familiar with. He immediately stood out in the passing crowd. We acknowledged each other with a nod, hesitating to engage in a traditional handshake, presumably each of us reminding ourselves of Covid-19. Daniel's hands showed the shaking movement typical of HD and he immediately hid them in his pockets. Despite half of our faces being covered with blue disposable surgical masks, I could tell that he responded with a smile. We agreed to walk to a coffee shop so we could talk. Daniel remarked, *'It is good to finally see you in person'.*

FIRST PERSON NARRATIVES

Daniel is in the early stages of HD. He lives in his family home and is independent in all aspects of daily activities. When asked about his life story, he was articulate and confidently expressed himself on the subject in spoken and written narratives. Daniel didn't choose to involve any of his family members in the research, he was however, happy for the HD advisor, Amanda Watts, to be involved the case. In addition to interviews, Daniel disclosed personal documents such his hospital letters and completed pages of a life story diary.

I presented Daniel's story in a 'first-person voice', as if he is speaking directly to the reader. This has been carefully constructed from my interpretation of the research data which included interview transcripts, observations, a day in a life diary and personal documents. This was a decision I felt strongly about, that voices tell a story (Frank, 2012) and that every analysis finds its own singular way (Frank, 2010).

In my review of HD literature, I found that it is very rare to hear a person with HD speak in their own voice (Mahmood, Law and Bombard, 2021), therefore I dedicate this space to the unheard voices of people with HD. Daniel referred to our conversations as the only time he had ever talked about his personal life experiences to anyone. After our first interview, Daniel said, *'I was thinking, this has been like, the only time I've ever talked to anyone really about my condition, even like, the day after the tests'.*

I also felt that our interaction permitted Daniel to share memories of his late mum. Given that the interviews were conducted shortly after his mum's funeral, it was as if Daniel felt the catharsis of talking for long periods without hesitancy. Reassuringly, my initial reservations about research engagement with Daniel had been put to rest. In my final interview with Daniel, he said, *'I really enjoyed doing all that, I hope I was useful for you'.*

I AM NOT HUNTINGTON'S

PART ONE: QUEST

If you want to know about Huntington's, this is not just about me. I grew up in a small family. It was just me, mum, dad, and my younger sister Cathy. I had a brilliant childhood, I felt lucky that way. My mum worked as a secretary and my dad was a mechanic. Because of dad's job we could only go away a few times a year. We all used to go *en masse* for a week in a holiday park. Sometimes, we would go abroad to places like Majorca, Turkey, and Cyprus. They were the good times.

But everything changed one day. I was 13 when mum and dad called me and my sister in their room. Mum asked us to sit down, she then showed us videos of very sick people and said this might happen to us in the future. It was too much for my dad, so he left the room. That was the first time I heard of Huntington's. I was in shock, the pictures of sick and dying people constantly played in my mind. I just couldn't get my head around it.

As soon as I turned 18, I decided I wanted to have the test. I was meant to go with someone to the hospital, but I kept making excuses like 'well, my auntie let me down'. Actually, I didn't tell anyone. I wanted to do it on my own. The idea was I would go take the test and tell everyone it was all clear and everything was a good laugh. On the day of the test, I took the bus to the hospital, but I didn't get off the bus. I stayed on the fucking bus. I wimped right out at the last minute. I thought maybe I was too young, maybe a little ignorant.

I was 26 when I finally took the test. This time I went with my auntie. We had the preparation for the tests, had four appointments and all the things that you've got to be ready for, to cope with it psychologically. On the day of my test result, I remembered we were waiting in a hospital room. They put us in a room full of kids' toys and ball ponds. The nurses were running late. Then the doctor came, and he hurriedly said: ' Oh listen, I'm really sorry it is bad news'. I remembered my auntie grabbing my hand saying: 'I'm really sorry'. I didn't know what to do. I couldn't get my head around it. I thought it will be a good result and that I am safe. Even after all the meetings, I guess you can never be prepared for this type of thing. For years I convinced myself that it was going to be a good test, and it turned out it wasn't.

When we walked out of that hospital, I asked my auntie a favour: 'Please don't tell anyone until I am ready'. She never told anyone. I kept my genetic result a secret. For ages, no one in the family knew. I think it was too much at the time, I was well getting into money worries, and I thought HD is gonna be the last straw. Also, mum was getting sick, and my grandad had probably enough guilt about my mum getting it and all that kind of thing. Everyone had enough to worry about, without me adding to it.

I remembered thinking, I might even go back to work soon after that morning, even though I booked the day off. I spent the rest of the day just walking around in places, I was trying to forget all about it. A few weeks later I decided to tell them at work. I used to work in a job centre as a team leader, our team had to ensure the roll out of new benefits and practices. Even though I didn't have the symptoms or anything, I thought that I am going to get worse, so it is best to put them in the picture. It felt a little bit weird when I told my manager, they sent me for a medical somewhere and to places where they sent everyone for their benefit assessments. It was funny because I have been sent to places where I work. The doctor who saw me didn't even know what HD was. He kept on asking me 'How's your dad?' for some reason he thought only men could get it. It was a bit weird that I was explaining more to him about HD, more than he knew. After that, it was all forgotten about.

I carried on working for a couple of years, but work was getting on top of me. Everything they were asking me to do was just too much. I thought, I am going to have a break before I get mad. I had one of those letters from occupational health when Cathy saw them. My sister thought something was wrong like I wasn't going to work. I told her I have been in work and those letters were because I had HD. She then dragged me to see mum and dad and I had to tell them what was happening. I felt funny explaining it to them, for ages they didn't know I had HD and I hadn't really thought about all that.

HD SECRETS: A FUNNY FAMILY THING

Obviously, I got HD from my mum, but she didn't know she had HD when she gave birth to me and my sister. Back then there were no blood tests in the hospitals like they have now. She got the HD from her mum, my Nan, but I don't really know the details on this. Our family wasn't really sure what was happening with my Nan at the time. They thought my Nan was just a bit mad and they put her in a psychiatric hospital. She died in a mental asylum. My mum was still young when her mum died. We found out that my Nan had HD when some people in America asked grandad if they could have my Nan's brain for research purposes. My grandad didn't know much about it either whatever it was, he said yes.

So, HD is a bit of a funny family kind of thing. It was only later on when I found out that others in the family might have it too. Some family members that I didn't know existed. Like, one day on the bus my auntie told me, 'Oh, I am going to visit my sister in the hospital', and I went on saying 'I hope she's alright anyway.' Then I realised my auntie's sister is my mum's sister too, and she had HD; my other auntie that I haven't met. My mum and her two sisters used to go to another city to see this doctor. Because one of my aunties she was really shaky, there was something wrong with her neck and her head was all over the place. People thought she had HD but it turned out she didn't have it. Nobody really expected that my mum would have it. She was the strong one in the family.

Back in the days when mum and dad used to go to the HD meetings, there were people there from the wider family too. So, definitely someone there had it. It's just one of those things that families don't speak about. When my sister took the test, she also kept it a secret. We only learned about it when she had the 'all clear'. She's okay now and so we are alright on that side I think. It's just me now [laugh].

PART TWO: OUT OF CHAOS

THE ACCIDENTAL CARER

It must have been about seven years ago when I left my job working in that job centre. My dad was still working, and mum was on her own in the house. When I had a break from work, I thought I will help them out for a couple of weeks as my mum was getting unsettled. I came back home to live with them, mum was made up having me there. She settled right down. Then it sort of fell into place when I stayed there. I looked after her. I became her main carer kind of thing.

Even though it was hard work, it just made me a happy person. I have always been a happy person, but work had been dragging me down. Coming back home was great, it allowed my dad to stay at work until he retired. My dad didn't have to worry because he knew I was at home with mum. I could ring him if there were any problems. It suited everyone and it worked really well. People used to see me and say: 'God, you look so happy', because I was happy. Everyone was also saying, they couldn't believe how well my mum looks after six months of me being there. You know, that was really cheering me on.

I often say without me being back at home, my dad wouldn't have been able to work in the last six years until he was 65. It suited everyone perfectly. My mum was happy as she liked a routine. Although my mum couldn't say anything, she liked having the same carers coming over to wash her hair or something. If this wasn't happening, she'd become really unsettled. We sort of made a pattern for the day and on the weekends, we would take her out in her wheelchair. She liked going out but it got her really tired.

As my mum's carer, I went to carers' events. I didn't think I'd ever do stuff like that. We went to the meetings a couple of times that had been organised by the council. There was this woman from the council who came out together with the social worker and she filled a form. She had never seen me before, but she was asking questions, like how happy I am, how often I think about the future and so on. Then one day, this woman rang me up and said she was worried because I scored really low on the form. She then sorted out some carers' vouchers which we could use and people to look after my mum so that my dad and I could get out and have a bit of time together. The carers' support network around us was brilliant, I have been to a few days out in cities and seaside towns. I had never gone to anything like these things before because I was worried about leaving my mum to people who didn't know her. These things were good, you can stay at certain hotels for free or do loads of things like Zumba, cooking and relaxation classes. It was really nice. It just helps you get out of the house and have a nice little day off. A day without really thinking much about a lot of things.

A part of me still thinks this all happened by accident. Although it sort of happened by accident these things happen for a reason. I looked after my mum until the day she died. It wasn't easy but it was probably the best thing I have ever done.

MUM

Everyone sort of says that my mum started to show the HD signs probably on her 50th birthday. We had a big party for her, and people noticed little behavioural patterns that started changing. As I say, everyone in the party was a bit taken aback. I never noticed it because I saw her every day. I remembered my mum went through loads of phases, like she was smoking her cigarettes and nearly set the house on fire. My mum loved a cigarette, there were times she wanted to smoke in her room, but we had to take all her cigarettes away from her because she was shaking and all that, so it wasn't a great combination. Also, at the time, her bedroom was still upstairs, she wanted to walk down the stairs by herself. I'd be behind her but she'd show how strong she was, she wanted to do it by herself. We eventually had to sit her down and explain to her that she might fall. From that moment, we moved her bedroom downstairs.

Even when mum started to lose her speech, she was still able to tell people if she didn't want something. Even when she got referred for a feeding tube in the hospital. She was losing loads of weight, so we went to see this woman we had been seeing for a couple of years. The woman said, 'Do you want this feeding tube?' my mum folded her arms and just went 'No!', and the woman went, 'Well, that answers that'. So, I say we knew exactly when my mum didn't want something, she would cross her arms to indicate 'I'm angry' and 'I don't want it'. But she kept on losing weight and eventually got the feeding tube. This should have been a straightforward operation, but she stayed in the hospital for two months.

My mum needed extra care at home, sometimes she would get agitated. Sometimes, she wanted to go somewhere, or she would stay in the room with us and watch telly. But a lot of times she just wanted to go to bed. If she wasn't getting her own way, she would like tip her chair or a couple of times she nearly fell out of her chair. She would lean so much out of her chair that we would have to make sure someone is there all the time sitting next to her so that she won't fall. You had to be there for her all the time. My aunties didn't come as much, they found her too hard especially when my mum started to get really sick. They stopped coming altogether. When they used to come, they would talk to her like a kid. I often said, 'That's not how you do it'. My aunty would come and say, 'Arre you..okayyyyy?' and I would think, ' Why are you talking like that, you talk to people with HD just like someone normal. People should talk and treat people with HD the same. They often asked me in front of mum, 'How's your mum?' and I'd reply, 'Well, she is there, ask her'.

Even when mum couldn't talk anymore, she would still be able to communicate in some way. There were signals like flicking her eyes when she was saying yes. They were the little things like raising the eyebrows. You have to watch these little things. Often, I asked mum if she wanted to go to bed, she would grab my hand a bit tighter to tell me, 'Yeah, I want to go to bed'. When my mum's carers came twice a week, the girls would bath her, wash her hair and she loved that. She loved getting spoilt. The girls would say to mum: 'can you help?' and she would. Even if there's one person helping her in bed, she would try to turn, and try to lift, she would still help people. The hardest thing is trying to explain to other people that she's

still my mum, just because she can't talk back doesn't mean she couldn't do anything or understand what other people are saying.

It was in January 2020, when she was really weak, she had a chest infection that she went into hospital. The nurses were building her strength up and they also found out she had a hiatus hernia. So, they couldn't put the feeding tube in and had to change to a rig type to secure it properly. This was because her stomach was higher up because of the hernia. There were different nurses and doctors who got involved, they were telling us in the hospital they wouldn't resuscitate her. There was a chance she wouldn't come through this. She had been through so much and she had just lost so much weight. But before she went into the hospital, I was still giving her dinner, like blended in a soup type of thing and a soft pudding like a trifle. She was starting to eat them all. But I noticed during mealtimes, she was coughing a lot. She also got frequent chest infections. Then all this Covid virus thing was just starting to happen, and the last place we wanted her to go was in the hospital. We really didn't want her to go. As you know, we had to do it in the end, then one thing led to another. What was supposedly a week in the hospital led to eight weeks, and we eventually got her back home in March. When my mum was home, we were getting used to feeding her on the PEG tube, but then she got weak again. From the time she came out of the hospital, she was having fevers and infections.

We got the doctor out to see my mum at home. He literally just went 'Oh, I think she might have Covid' and then walked away. With HD, you can't fight back from a virus. We didn't really know what to do, we didn't know if we should we give her food or something. That Friday afternoon, a district nurse came and said: 'She is not going to make it through the night'. The carers that came with mum when she came out of the hospital heard about it and they were all in tears. I remember I said, 'You know what my mum's is like, she'll be here on Monday'. When the carers came back on Monday, there was my mum wide awake in bed waiting for them. My mum was so strong, and whenever I'd think, 'It is not long now, fight back, fight back', the next day she'd be okay, she would be sitting with us in the front room. Every night after, my dad and I would make sure someone was awake there with her. Even though she wasn't opening her eyes, we just held her hand and all that kind of thing. Ten days later my mum died.

Right up to the end, my mum was just strong. Even the priest who did the funeral said, 'everyone says the same word. All I am hearing is strong, how strong my mum was, she was a fighter. My mum was just amazing, everyone was saying in the funeral how well we looked after her. All we did for her wasn't easy, but she was just so strong no matter what they threw at her. She had gone through a lot, even a cancer scare, but they later found it was an ulcer. After the operations, and the many other things she had taken out, she just came back stronger and stronger. My mum was dead strong with all of it, she has been a great example of how to be strong through all these kinds of things.

PART THREE: RESIGNATION, TO WHAT LIES AHEAD

MY MOTHER'S DISEASE IS MINE

With having HD I kind of thought, 'Okay, get on with it'. I got involved with the HD program in the hospital. I went to see the consultants in the clinic, and they began to check me every year. I would go through some tests to see how my HD was progressing, so they could keep an eye on me. It is just once a year, and nothing really changes. Sometimes, I think it is pointless.

I like going to these types of things like I have done a couple of times where they bring new GPs in. The nurse in the hospital would ask me if I could be 'the patient kind of thing'. The tutors were there, and they would look at my movements and I would show them what I do. It is good to teach people about HD.

My mum never had that sort of regular care from the hospital, like no one would check her arms or legs making sure she was okay. We had a nurse who would check once in a while, but no one actually checked my mum properly kind of thing. We have been going to our family GP for many years. They were brilliant, the people who work there know our family. If we needed an emergency GP appointment, I never saw any hesitation from them. We are lucky as the same surgery has been run by the same family for some time and the same doctors have been there for ages. It's good that way, so I don't have to explain to new people about HD all the time. Aside from that, there's no other NHS people who are involved.

I was also supposed to be in a research drug trial where they put something on your spinal fluid but that got cancelled because of all this Covid that happened. At the moment, all they are doing in the hospital is about Covid. I applied again for another research trial, they have accepted me, and they will get back to me once they start.

HD MADE ME A BETTER PERSON

I am single and nothing has ever really happened, didn't get engaged or anything like that. It's just me and my dad now. I do most things on my own at the moment. I do most of household work, so my dad doesn't have to do it. I'm worried about him as he has been struggling with his breathing lately. He has COPD. I'm trying to look after him.

I still go to the bank and all that, although, I need to have a word with them as I noticed my signature has been changing. I can't remember at times how to sign my name. With what has happened recently, this made me think I need to get a hold on these things. I'm a bit up the wall with online banking and other things, with Zoom and all that. At the moment, I find it harder everything going online. But I'm finding ways to make it easier for me in the future. I think I'll get used to it; it just takes me a while.

I also noticed I struggle with my balance lately. I started walking weird and had a few slips. The last time I went in the clinic for my HD assessment, I had difficulty with the baby steps: when you put one foot in front of another; like I am bouncing off the walls a bit there. Also, when they asked me to repeat three patterns with my hand, I couldn't remember the third one. The main thing at the minute is my spatial

awareness, just looking at things and finding them. I try to grab a book and end up not picking it up or knocking it off. That's the worst thing as I get a bit frustrated with myself. I've always had really good coordination. Whenever I've done this in the past, I've always caught things before they hit the ground and now I can't, that doesn't happen anymore.

I am learning how to concentrate more and get a better memory. But that's the thing, I don't know how much of my memory is normal and how much of it is HD. There are times that I go back to the fridge, and I can still see it open, that could only be me. I am trying to work on little ways of improving my concentration, to make sure that I do concentrate on everything. It is the little things like picking up a cup on the side, because if I get it wrong the chances are I will knock that off and I am gonna break the cup. The main thing is, I think HD has changed me a bit. Although, I am still kind of the same.

HD made me a better person. It definitely made me appreciate a lot of things like family. The thing about HD is that there is so much of it that is so unpredictable. People don't really know for certain, like what age it starts. It comes on at different times, quickly in some and slowly in others. I just thought, 'Okay, well whatever happens, you know we're gonna make sure we enjoy it kind of thing'.

The other thing that I have been a bit worried about is that I might lose the plot a little bit. But I think I'm coping alright with it, you know. I waited a long time for the bereavement counselling, but I think it's not gonna happen now. They told me they will get back when all this Covid dies down, I haven't heard anything since.

Life has been up and down. But lately, I keep hearing noises when I'm in the front room. Like with my mum, I can hear her voice and there were sounds of other people in the front room, and I run in there but then there's no one there in the room. I sort of think I have got a handle on everything but something weird happens, but we're all okay...

EPILOGUE

In our final interview, I met up with Daniel again to personally thank him for his research participation. As I expected he was there early at our meeting place before our agreed time, and I had come early so he didn't have to wait long. This time we shook hands, walked to the same café, and took our masks off once we sat down at our table.

'It's good to see you again,' I said, *'and thank you for everything'.* I presented what I wrote about him and invited his opinion on the idea that I wanted to present his story in his own voice. He agreed to this and seemed pleased. Daniel replied,*' Thank you for listening to me, and I hope I didn't talk too much'.*

Daniel was further pleased to inform me that he had received a pre-screening information sheet inviting him to participate in a new HD clinical trial to commence in the summer of 2022. He clumsily scuffled in his pockets to show me the hospital letter with the details of the research; a randomised double blind,

placebo-controlled study administering intrathecal treatment for patients with early HD. Daniel sounded enthusiastic about getting involved with research trials, despite the unsuccessful results of the previous HD drugs (Tabrizi et al., 2022). He said that ever since he had heard about drug experiments in HD, *'I always wanted to do it'*. I asked Daniel why he was so eager to participate, to which he stoically replied, '*To be honest, I think nothing is going to ever happen, but I need to do what I can. We all live in hope'.*

After a couple of hours, when we had nothing else left to say to each other, the time had come to part ways. Daniel's final words to me were, '*It's hard to explain, you know… I didn't want to be the Huntington's person; I just want to be me'.*

CHAPTER 6: DISCUSSION

This study provides insights into how people with HD narratively construct their illness experience at different stages of the disease. Jen, Martin, and Daniel's illness narratives conformed to Frank's (1995) storylines of Quest and Chaos. In addition, the study identified a novel storyline of Resignation, because Restitution has 'receded farthest into the background of the stories of the chronically ill (1998 p.201), and when it is no longer attainable other storylines emerged (Frank 2012).

The HD storylines (Figure 6.1) as exemplified by the study participants exposed the wider impact of the illness. While Jen, Martin and Daniel elucidate the meaning of living with HD from within a personal context, they also illuminate the salient experiences for their families. Their stories revealed the broader social effect and the complex issues surrounding this rare, often misunderstood hereditary illness. Despite the improvements in scientific research and advances in medicine, people with HD and their families, as illustrated by these stories, were left behind. These stories have both a general relevance and some specific implications for clinical practice, and this will be explored at the end of this chapter.

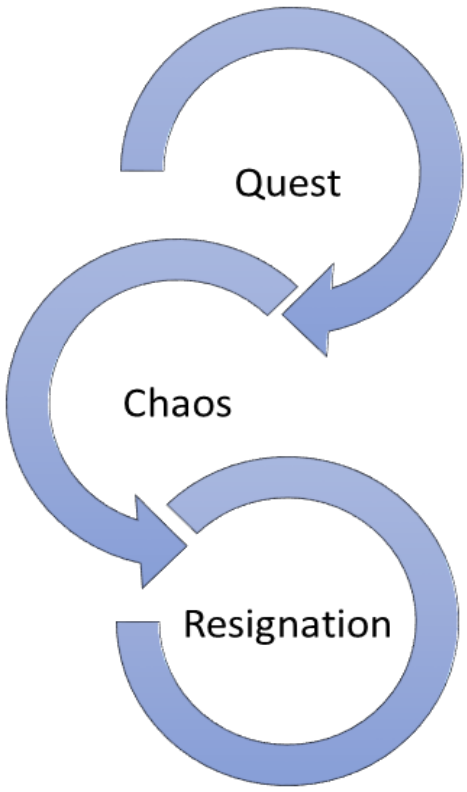


Figure 25. HD Storylines

BROADENING FRANK'S ILLNESS STORYLINES

Illness narratives are widely used to better understand the importance of the patient experience (Kleinman, 1988; Sakalys, 2003). Frank (1995) and other scholars (Robinson, 1990; Hydén, 1997; Bury, 2001; Sparkes, 2005; Murray, 2008) have used typologies or storylines to encourage closer attention to the nuanced experiences described in, and the distinctive features in the stories of the ill. In this study, in addition to exploring the HD experience and presenting them in storied data, model storylines across the three narratives cases were identified. Listening to illness narratives can often be problematic because they lack coherence, but using the storylines as listening devices can help weave the multifaceted narrative threads (Frank, 1998a).

The narratives of Jen, Martin and Daniel revealed the uniqueness of their personal illness experience. Although it is not possible to homogenise their experience, they can be 'representative in that uniqueness' (Frank 2010 p. 116). The HD storylines 'underlie the plots and tensions 'of their stories (Frank 1995 p.75) and have provided a 'general unifying view' (p.76). In the contemporary literature that concerns itself with illness narratives, one can find frequent citations of Frank's storylines of Quest, Chaos and Restitution. This typology has been echoed in many illness narratives, including those that concern chronic diseases and long Covid-19 (Appendix B, Table 4). One of the novel contributions of this study is that for the first time, Frank's narrative approach has been used in understanding the illness experience of people with HD.

Critics of Frank's approach have argued that typologies can limit the narrative genre (Atkinson, 1997; Hyvärinen, 2008). Atkinson (2009) critiqued that although a storyline is useful as a starting point, the focus is 'too restricted to an array of narrative functions' (p.199). Frank (1995) acknowledged that the illness experience does not conform exclusively to one type, and that different storylines can be told repeatedly or interchangeably at different moments on the illness journey. The risk of a general unifying view is that it depreciates the uniqueness of the individual illness experience, and Frank (1995) made it clear that 'other types of narratives can and should be proposed' (p.76).

Therefore, in addition to Frank's (1995) established illness typology of Quest, Chaos and Restitution, this study proposes the novel storyline of Resignation. Resignation, as defined in the Oxford Dictionary, is 'the quality of being willing to accept a difficult or unpleasant situation that cannot be changed'. Resignation was highlighted by the HD participants themselves as to how they see a future that cannot be changed with the inexorable disease progression. However, it is important to point out that their resignation was not synonymous with despair, loss of hope, nor despondency. Rather resignation was an act of resolution in response to their situation and their disease. Martin expressed his resignation poignantly, as an intention to 'learn to live with what I've got'. Daniel believed that his illness would take over his life and was resigned to 'what lay ahead with the HD'. Jane was resigned to live her remaining days in the nursing home with 'I am happy here'. Similarly, Martin shared the intention 'to die happy'. Their resignation narratives implied they were no longer fighting against HD but living, accepting, and having a new relationship with their selves and their illness.

WHEN RESTITUTION IS NO LONGER ATTAINABLE

Findings from the study both agree and disagree with Frank's claim on the restitution narrative. Restitution was the preferred illness storyline, especially with HD caregivers as this reflects their natural desire for HD sufferers to get well. This also aligns with the predominant biomedical model of health to implement treatment and provide care to relieve disease and suffering (Kleinman, 1988; Hydén, 1997). This institutional model strongly influences 'how illness is to be told' (Frank 1995, p. 78), with the prevailing agency of the medical community (Donnelly, 2021). Frank claimed that restitution dominates the stories of 'the recently ill' (p.77), but they 'no longer work when the person is dying or when the impairment will remain chronic' (p.94), and when restitution is absent, a 'narrative wreckage' will reveal itself (p.94).

While Frank's Restitution storyline was pertinent to the narratives of families and HCPs, it was largely absent in the narratives of HD individuals and was replaced by 'Resignation'. Firstly, in the HD participant's diagnosis stories, and even after receiving gene-positive confirmation, they knew from the experiences of their relatives and families that their future could not be restored, and in Daniel's words it was 'only a matter of time'. Secondly, as observed in the participants' accounts of their earlier experiences, they defied the restitution narrative of 'I'm sick, but tomorrow I'll be healthy again (Frank 1995, p77)'. This is rather aptly illustrated in the article title of a recent review of HD lived experiences by Mahmood et al (2021) which states, 'I have to start learning how to live with being sick' (p.1). Jen, Martin, and Daniel knew that with HD, the disease can only get worse and eventually the illness would take over their lives. In Martin's words, 'be ready for it.' In the absence of Restitution, a different storyline emerged, which is Resignation.

RESIGNATION

The Resignation storylines discussed in this thesis stand in marked contrast to the way in which the participants in Charmaz's (1991) study on chronic illness viewed their illness, namely as 'something to struggle against or to defeat' (p.13). Restitution is mostly absent in the HD narratives as the illness progression cannot be halted, cannot be cured despite the advances in research, medicine, and technology. Once symptoms develop, the condition can only be managed and the old self (Frank 1995) can never be restored (Moulton, Hopkins and Bevan-Jones, 2014; Petersén and Weydt, 2019). The HD participants were seen to slowly accept, based on personal experience, that they eventually 'will become sick'(Mahmood, Law and Bombard, 2021), and their mind and body will fail (Huntington, 1872). Martin expressed his resignation in such terms: 'I'm going to go a lot worse than what I am'.

The participants' resignation can be viewed as an adaption to the illness. Charmaz (1995) proposed that a serious chronic illness, like HD, 'undermines the unity of body and self and forces identity change' (p. 657). McAdams and McLean (2013) explored this narrative identity change through adaptation, and they identified the redemptive theme that people use to turn the negative events of suffering into a positive resolution. In chronic illness, adaptation is a means to resolve the disruptions caused by the disease (Williams, 2000; McAdams and McLean, 2013; Roger et al., 2014; Habermas and Köber, 2015). The stories

of Jen, Martin, and Daniel showed that they have adapted to and accepted HD by creating narratives around their Resignation. In common with HD literature that has provided descriptive themes of 'adaptation' and 'coping' (Helder et al., 2002; Etchegary, 2009; Roscoe et al., 2009; Arran, Craufurd and Simpson, 2014; Wauters and Van Hoyweghen, 2021), these resignation stories acknowledge that persistent HD challenges will continue to exist, but rather than a struggle against it, they struggle with it.

The Resignation storyline reminded me of a passage from Victor Frankl's (1985) book, In *Man's Search for Meaning* (1985), in which he wrote, 'When we are no longer able to change a situation - just think of an incurable disease… we are challenged to change ourselves' (p.112). Resignation is not tantamount to giving up, but to an acceptance of the limits of the body and the situation, and to a surrender to the outcome of living with HD. Jen, Martin, and Daniel changed Restitution to Resignation because they have no power over their disease nor the illness' foreseeable trajectory. This change can also be likened to what Charmaz (1995) described as 'relinquishing control over illness and by flowing experience of it' (p.657). Jen, Martin, and Daniel have changed their relationship with their illness, repaired the disruptions caused by it, and altered their outlook to accommodate and accept HD in their lives.

QUEST

Alongside Resignation, Arthur Frank's storylines of Quest and Chaos permeated the participants stories. Quest is a pervasive narrative found when participants sought information and care management to overcome the disruptions of their illness in their everyday lives. But the most powerful appearance of the Quest narrative can be seen in the participants' attempt to find resolution upon receiving their diagnosis, and to uncover the HD secrets within their family.

DIAGNOSIS AS QUEST

All stories, including stories about illness, have a beginning. When I first asked Jen, Martin and Daniel to tell me their story of living with HD, their opening narratives were about how they received their diagnosis and how they revealed this to others. The disclosure of their HD status was the beginning of their 'real' experience of living with HD. These stories centred around diagnosis and genetic tests have dominated HD qualitative literature (Audulv, Packer and Versnel, 2014; Mahmood, Law and Bombard, 2021). Experiences of HD diagnosis have presented varied themes (Schwartz, 2010; Etchegary, 2011b; Hagberg, Bui and Winnberg, 2011; Hagen, 2018) but what the accounts given by Jen, Martin and Daniel revealed was their Quest for the personal meaning of having HD. Initially, Daniel hid his diagnosis from his family for the main reason that he didn't want to burden them. However, he claimed that after revealing the truth to his mum and dad, he took actions that 'made him a better person'. Ironically, Martin and his wife, Pamela welcomed the HD diagnosis. The couple resolved their marital troubles and strengthened their vows to face a future with HD in it. Poignantly, Pamela stated that 'Huntington's saved our marriage'. Jen, after revealing her diagnosis to her children, was left with concerns for the future, in particular the risk of passing the disease on to her children. She was concerned about the risk of passing the disease on to her children. However,

her illness somehow brought her family together. When she could no longer find strength in herself and the illness overcame her, it was her children who stepped up to support her. For Jen, Martin and Daniel, the HD diagnosis had been contextualised, and could be viewed within the broader context of its social meaning, taking in the perspectives of the people who cared for them.

The participants' stories help us better understand researchers' previous suggestion that the period of testing and receiving diagnosis is a critical moment in the HD experience (Broadstock, Michie and Marteau, 2000; Crozier, Robertson and Dale, 2015). Indeed, experiences of HD diagnosis, as with other neurodegenerative (Paulsen et al., 2013; Peek, 2017) and genetic conditions (McAllister et al., 2007) are critical, as revelations of an incurable fatal illness can be life changing. However, Peek (2017) in her study of 37 people with Parkinson's Disease (PD) highlighted the shortcomings of HCPs who handled these diagnostic encounters and that the 'human significance' (p.35) was dismissed. HD and PD are both progressive movement disorders, and in each case the disclosure of diagnosis can be traumatic, having a significant impact on both the psychological and social experience of the affected individual (Schwartz, 2010; Peek, 2017). Alarmingly, the highest rates of HD depression and suicide have been recorded during the period of diagnosis (Paulsen et al., 2005; Kachian et al., 2019). This study adds weight to existing HD literature in emphasising the imperative need for support in receiving diagnosis with particular attention to the HD patients' social situations and environments (Richards, 2004; Schwartz, 2010; Ghosh and Tabrizi, 2018; Smedley and Coulson, 2021). This study further highlighted that support should also be extended to families, such as their spouses and children due to the distinctive impact of HD on the family system related to the disease genetic transmission.

The participant's Quest stories around their diagnosis illustrated the lack of appropriate care interventions or support for their psychological and emotional burdens after receiving the test results. Despite best practice guidelines existing for predictive testing (MacLeod et al., 2013) and diagnostic testing (Craufurd et al., 2015), and the availability of both genetic protocols (Groves, 2017; Quaid, 2017), and recommendations for counselling (Semaka and Hayden, 2014; Stopford et al., 2020), Jen, Martin and Daniel's experiences suggest that health services were limited, and that provisions vary. They claimed to have not received further follow-up support from HCPs after receiving their diagnosis. In common with other genetic and rare diseases (Morgan et al., 2014), in the absence of established and readily available support, the participants turned to the internet, online forums and support groups (Smedley and Coulson, 2021), which led them to the HDA charity. The diagnosis stories imply the need for better psychological access, care and treatment for HD patients and families (Broadstock, Michie and Marteau, 2000; McAllister et al., 2007; Zarotti et al., 2020), and that this support should continue beyond the disclosure of diagnosis (Decruyenaere et al., 2003; Andersson et al., 2016; Nance, 2017).

QUEST TO UNCOVER FAMILY SECRETS

The HD participants sought to uncover the family secrets that had been kept for many generations. As can be seen in other accounts found in HD literature, they shared stories about distant relatives, 'riddled with

vague talks' about a 'schizophrenic grandmother confined in insane asylums' (Bird 2019 p.51). They spoke of relatives who were marginalised and vilified (Wexler, 2002) as a consequence of their psychiatric behaviour (Halpin, 2011). Like Bird (2019) who collected stories from generations of HD families for over forty years, Jen, Martin and Daniel's stories compel us to better understand how individuals and families live with the inherited risk of serious mental disorders.

The HD secrets persisted in the participant's stories in a similar way to what was described in HD literature (McCormick, Korf and Wexler, 1992; Wexler, 1996; Sobel and Cowan, 2000; Forrest Keenan et al., 2009; Mantell, 2010; Schwartz, 2010; Bird, 2019; Wauters and Van Hoyweghen, 2021). They have been found to be ubiquitous in diagnosis stories and in family histories, and as Wexler (1995, p. xvii) has written, 'Secrets, moreover, especially so dramatic a secret as Huntington's, may form part of a family's emotional inheritance, a psychological legacy handed down along with the family Bible, affecting every aspect of family life for generations'

Frank (1995) observed that most published illness narratives are structured as a quest. Illness is lived as a quest (Frank 1998, 2002). Frank (1995) ascertains that, 'Illness is the occasion of a journey that becomes a quest (p.115)' and goes on to say that 'what is quested for may never be wholly clear, but the quest is defined by the ill person's belief that something is to be gained through the experience'. This has a strong resonance with the participant's stories which tend to assert that despite their diagnosis of a terminal illness, HD has brought a positive effect on their lives, such that in Martin's wife, Pamela's words, 'HD saved our marriage' and for Daniel, 'HD made me a better person'.

Frank (1998) further states that 'Quest stories are about illness leading to new insights' (p. 203), and that they pursue 'what can be learned and how this lesson can be passed on', (p.204). In my final interviews, I asked the question, 'how would they feel when I shared their personal stories with others?' Their response was altruistic in the way that they hope something could be learned about living with HD. I agree with Frank (1998) that the most moving stories are those, like Jen, Martin, and Daniel's illness experiences, that are lived and told by the ill. In finding reconciliation with HD, of 'what can be reclaimed of life' (Frank 1998, p. 204). I borrowed Pamela's words that 'life with HD is not all doom and gloom', the participants' Quest stories show us how 'they too could be living' (Frank 1998, p. 33).

CHAOS

The illness storylines of Chaos were embedded in Jen, Martin, and Daniel's accounts of episodes of depression, suicidal ideation, and metaphors of breakdowns. When the participants spoke of their Chaos, Jen's children felt 'no one listened' to them. Likewise, when Pamela sought help with her husband's deteriorating mental health, she received a disheartening response from HCPs that 'there's nothing we can do'. Daniel felt after talking to a doctor about his condition that 'it was all forgotten about.' Similarly, in Timothy and Marjorie's appeal to help their despondent mother, they felt 'dismissed' because 'all they see is this alcoholic aggressive woman who refuse [sic] to engage with them'. Their breakdowns reveal their

feelings of helplessness. In a sombre recollection when Jen was confined in a psychiatric hospital, heavily sedated and looking emaciated, Marjorie remarked 'no one cared.'

The experience of chaos extended to HD families, and Timothy and Marjorie recounted growing up with 'chaos everywhere'. Pamela also had her breakdown story as she recalled being unable to bear the aggressive behaviour of her husband, leading the couple to consider ending their marriage. Frank (1995) explains that when in chaos, 'troubles go all the way down to bottomless depths. What can only be told begins to suggest all that is wrong' (p.99). In the participants' stories, Chaos started to emerge before the official HD diagnosis. While family members first noted the physical changes with the emergence of erratic movements, it was the behavioural changes they were mostly concerned about. While the physical symptoms of HD are 'disabling', it is the neuropsychiatric symptoms that are most devastating to individuals and families (Craufurd and Snowden, 2002; Williams et al., 2009b; Halpin, 2011; Schumacher-Kuiper et al., 2021).

Painting in broad strokes, the participants' Chaos stories were prompted by unmet health care needs. This was largely because HCPs were not able to understand their needs. Moreover, Jen, Martin and Daniel had each at one point 'fallen through the net' (Shakespeare and Anderson, 1993, p.19), with their complex neurological, physical and psychiatric symptoms not fitting neatly into the available care systems. The HD symptoms cross the boundaries between the disciplines of neurology and psychiatry (Ehret et al., 2007; Halpin, 2011; McAllister et al., 2021), to such an extent that in Pamela's words, they've been 'passed around' between these two services. Likewise, Daniel was seen by a doctor who he felt he was 'educating more about HD'. Similarly, health provisions, according to Jen's social worker, Poppy, are still based on 'old' delineated 'medical and social models of health', and Poppy got frustrated with the responses from her colleagues who said, 'well this is the condition, these are the behaviours and there's nothing we can do about it'. Moreover, given the structure of British social services departments, Poppy suggested that 'our mental health teams are split so you've got the specialist adult mental health team which deal predominantly with people with dementia and then you've got the assessment and treatment which is working with people with some mental health problems and they're usually of working age and those approaches are very different'. Given their range of physical and mental health symptoms, people with HD necessarily need support from both disciplines. For Timothy, the current health, and social services 'do not communicate with each other' and as a family member he 'felt at a loss,' in that if a person with HD doesn't have the right support around them, 'how will they be able to get around this quagmire system?'.

Chaos stories were not easy to listen to, as they do not fit the traditional storytelling format that offers a clear resolution (Donnelly, 2021) and this contrasts with the health professional's preferred narrative of Restitution (Frank, 1998a). The participants echoed the findings of studies on HD family caregivers that many health professionals do not have any real understanding of the disease (Nance, 2007; Skirton et al., 2010; Wilson, Pollock and Aubeeluck, 2014; Edmondson and Goodman, 2017; Anderson et al., 2019) because they failed to really listen to what is important for the person with HD and their families. Wilson et

al (2014) observed that knowledge of how to provide care for people with HD is limited and perhaps one of the key steps to overcome this is to truly listen to people's illness experience (Lang, Floyd and Beine, 2000). Consistent research suggests that people with HD and their families do not receive the health care and support they need (Dawson et al., 2004; Soltysiak, Gardiner and Skirton, 2008; Skirton et al., 2010; Etchegary, 2011a; van Walsem et al., 2017), which negatively impacts their quality of life (Helder et al., 2001; Aubeeluck, Wilson and Stupple, 2011; Ho, Hocaoglu and European Huntington's Disease Network Quality of Life Working, 2011; Anderson et al., 2019; Aubeeluck et al., 2019).

The Chaos stories were 'anxiety provoking' (Frank, 1995 p. 97) and recalling them triggered emotional responses in most participants. In my interviews, there were instances where we had to pause the interview, because participants cried, and were upset upon recalling these experiences. However, the participants chose to continue in the telling of their experiences. Their long engagement in the study with repeated interviews allowed them to reveal the storylines of Chaos in their own terms. As Frank (1995) argued 'to turn chaos into a verbal story is to have reflective grasp of it' (p.98). The study findings support Frank's claim that chaos can only be described when the participants are able to recount their experiences retrospectively, contemplate them and achieve distance from them. This was also made possible when the participants read, discussed, and reviewed the interview transcripts and engaged with my analysis of their core stories. They consequently turned their lived chaos turn into reflection (Frank 2005).

'THE OTHER' STORIES

What was apparent in my interactions, particularly with Jen and Martin, is their acquiescence with the 'other stories'. *The other* refers to the stories of spouses and children. Jen encouraged me to listen to the accounts of her children, Timothy and Marjorie. In turn, her children involved Jen's social worker, Poppy. Martin strongly insisted on bringing his wife, Pamela. Although Daniel felt he didn't want to trouble his dad or his sister, he was favourable to soliciting the views of his HD advisor, whom his family rely on with any HD related concerns. *The other* narratives tell equally important stories in their own right. They reveal what Frank (2010) described as 'resonance' in storytelling, that 'one story calls forth another' (Frank 2005 p. 967), and the participants' stories echo other stories adding cogency to our understanding of the HD experience.

Moreover, Jen, Martin, and Daniel told stories that compared their own symptoms and experiences with their HD affected family members. Within their own stories, they narrate stories of their families, and here we find what Frank (2020) referred to as a 'nesting of stories and relationships' (p.2). Interview studies on HD (Helder et al., 2002; Hagberg, Bui and Winnberg, 2011; Booij et al., 2013; Williams et al., 2013; Hagen, 2018) and other genetic diseases (Kenen, Ardern-Jones and Eeles, 2003; Petersen, 2006; Werner-Lin and Gardner, 2009) have similar findings: that individuals tended to speak about a future constructed around family narratives. These existential struggles were noted by Klitzman (2009) that HD familial histories significantly affect the person's sense of identity. The participants have adopted what Leotoni (2006) referred to as the gaze of 'biomedical prediction', where they foresee 'how they will be' (p.151). The HD

participants had preordained their future by what they saw and have known from the experiences of their families. They believed that what is in their genes and their illness will determine them. Martin's story exemplified this determinism as he recursively compared his mum's mental health and the care treatments she received. He believed that 'shock therapy' is the 'only way' in the event he gets worse, despite his wife's observations that he had good outcomes with his 'tablets'.

Timothy, Marjorie, and Daniel echoed Sparbel et al's observations (2008) that growing up with HD parents meant 'watching and waiting' in 'the shadow of HD' (p.327). Listening to their stories reminded me of Roach's (2018) article about a young man as his 'future etched in his father's face' (p.1). It revealed how HD moves the illness experience from family narratives to the individual. Jen, Martin, Daniel and the HD children, Timothy and Marjorie largely narrated how they witnessed that HD clings to the bitter end in their parents, and relatives. Their family narratives intertwined with and assimilated into their own illness experience.

The recent international validation of an HD quality-of-life clinical tool for HD caregivers (Aubeeluck et al., 2019), is a recognition of how HD negatively impacts the health and wellbeing of HD family caregivers. Researchers have identified the unique HD family needs and recognised the particular importance of the experience of HD spouses (Kessler, 1993; Richards, 2004; Andersson et al., 2016), family caregivers (Williams et al., 2009b; Dale, Freire-Patino and Matthews, 2014; Domaradzki, 2015; Røthing, Malterud and Frich, 2015a; Parekh, Praetorius and Nordberg, 2017), and HD children (Williams et al., 2013; Kavanaugh, Noh and Studer, 2015; Kjoelaas, Tillerås and Feragen, 2020).The findings of this study resonate with Brouwer-Dudokdewit et al's (2002) study of six HD case series, which suggest that HD as a family disease interrupts the 'the family cycle' and 'forces its members to cope with disruptive events and untimely deaths' (p.689). While the latter study focused on the impact of genetic testing, the present study extends its findings that care and support for the HD affected families must be considered more deeply. Similarly, Rolland and Williams (2005) proposed a Family System Genetic Illness model to address the inherent psycho-social challenges of genetic disorders such as HD. People with HD and their family caregivers have an overall decrease in quality of life (Aubeeluck et al., 2019). The experiences illuminated in the narratives of HD families involved the study, together with the person with HD, they too have their burdens, and they seek to make sense of HD in their lives.

GENERAL IMPLICATIONS

The findings of the study were context, place, and time specific. Nonetheless some aspects of the research could be significant to other people living with, and affected by HD. This section will highlight the implications applicable to all those who provide HD care.

THE PERSON BEHIND THE STORY

Jen, Martin, and Daniel disclosed the burden of living and dealing with HD. Their stories remind us of personal struggles but also illuminate their coping strategies, their resilience, and their ability to identify

sources of support. They asserted their own values, sense of self and identity in the face of a disease that gradually takes away their physical and mental capacities. As in the case of HD, it has been observed that people living with a range of chronic illnesses experience the fundamental 'loss of self' (Charmaz, 1983 p. 168) in the face of 'biographical disruption' (Bury 1982. p. 167). Their illness experiences demonstrate an 'ongoing struggle' (Kaite et al 2015, p.1) as they 'grapple with control' (Mahmood et al, 2021 p. 12) to 'maintain a sense of self' (Roger et al, 2014 p.1) and 'adapt' (Telford et al, 2006, p. 457) to the capricious demands of their illness. In this study, the loss of control has been evident in the participants' Chaos stories, yet their Quest stories also show how they have become reconciled to the past and moved on to Resignation stories to accept and to live with a terminal illness. Nevertheless, Jen, Martin, and Daniel's Resignation does not signify a future lived with passivity. They call on us to see them as people, who are still capable; to understand, make informed choices, and take action in their lives. They wanted to be called by their names, communicated to, and treated as a whole person with their illness just a part of them. 'Never lose sight of the person', the words of a mentor colleague resonated as I recalled my first HD patient in a care home. He was a large, unkempt, apathetic middle-aged man with wild flailing movements that led to falls and displays of aggressive behaviour. In a similar fashion to Jen's social worker, Poppy, I was taken back by what I saw and what I read from his case notes. After hearing his personal story, his previous job and hobbies, love for animals and for his children, I no longer identified him with his illness. It was through his narratives that I was able to therapeutically engage with him, as we worked on his physiotherapy goals. Thus, knowing the person through their personal stories, the basis of the narrative approach utilised in this study, is one way to support them and 'see the person' behind their illness.

STORIES ECHO OTHER STORIES

The HD participant's stories convey that despite HD being a rare disease, if their families and carers are counted, the illness affects a considerable number of people The inescapable consequence of HD extends beyond the suffering of Jen, Martin, and Daniel. The 'ripple effect' (Schwartz, 2010; Kjoelaas, Tillerås and Feragen, 2020) of the illness experience is illuminated by their family members. Hearing other stories of 'the forgotten person of the Huntington disease family' (Kessler 1993, p.45) enables insight into the broader impact of HD. There is no doubt that education and public awareness about rare diseases such as HD are still needed. Most rare diseases are hereditary and chronic (von der Lippe, Diesen and Feragen, 2017), these diseases lack appropriate care treatments and research is limited to develop effective interventions for affected individuals (Gaasterland et al., 2019), often resulting in patients and families having a poor quality of life (Slade et al., 2018; Aubeeluck et al., 2019; Sokol et al., 2021). The National Service Framework (Department of Health 2011) recognised the need for service provisions to improve the quality of life of people living with long term neurological conditions and their family caregivers. However, as Wilson et al (2014) observed, the guidance in the UK refers to neurological conditions as a whole, and HD is rarely included, as the emphasis is placed on common conditions such as PD and MS. There remains a lack of evidence for clinicians to support the needs of people with rare diseases, such as HD. The implications of this study are that the patients and their families should be encouraged to share their own

stories, for they help us better understand the changes, disruptions and distinct needs that arise within family systems and social support. One way to help improve support for families is for these HD stories to come out of the shadows. Not only are narratives a valuable therapeutic means, but also useful in terms of research (Charon, 2012). Patients' stories 'can provide useful lessons, inspiration and guidance' (Wexler 2012, p.3), and their voices can help guide the direction of drug trials and therapies (Simpson et al., 2016), and suggest improvements in systems of care (Charon, 2001; van de Bovenkamp, Platenkamp and Bal, 2020).

THE USE OF STORIES FOR HEALTH AWARENESS

The stories of Jen, Martin and Daniel uncovered how personal stories can be used to highlight the need for health awareness, and with a particular emphasis on what is important for the person with HD and their family. Bingley et al (2006) reviewed narratives written since 1950 concerning people dying from cancer and other life-limiting diseases. Their findings illuminated that these sources of knowledge are important to people faced with chronic illness and when approaching death. Topics include the social and spiritual aspects of dying, alongside symptom control and issues of communication with medical staff. Molzhan et al (2019) found that narratives of people with advanced chronic diseases need to share and understand the stories of other people facing terminal diseases. In common with what Bingley et al (2006) and Molzhan et al (2009) observed, the participants in this study expressed their strong sense of purpose in sharing their stories.

Recently, the Huntington's Disease Alliance (2021) of England and Wales, an organisation composed of HD charities and funded by the pharmaceutical company, Roche, launched a public awareness campaign that centred on family stories (Appendix 0). Similarly, some NHS trusts have utilised patient stories to provide education and to influence the structure of service provisions (Wilcock et al., 2003; Condon, 2019); and evidence exists to suggest the potential of narratives to influence health-policy making (Fadlallah et al., 2019). The HD stories in this study and the many others published elsewhere are invaluable resources. Avenues that allow the production and dissemination of stories that explore a rare, complex, incurable disease like HD need to proliferate. Illness stories can be accessible vehicles to connect communities (Frank, 2006; van de Bovenkamp, Platenkamp and Bal, 2020) and they have the potential to fulfil a variety of purposes: to inform, to remind of invisible needs, to educate others, to stir emotions, to gain sympathy or to persuade others to action (Kleinman, 1988; Charmaz, 1991; Frank, 1995; Mishler, 2005; Charon, 2012; Sacks, 2014). Narrative methods are increasingly relevant in the current climate of public and patient involvement strategies (Gregory, 2010) and can help improve health services (Condon, 2019). In the absence of cure, these stories can help foster 'engagement with the HD family community in care research and health care improvements' and this perhaps can be a 'rate-limiting step in the search for better care' (Edmondson and Goodman, p.177).

CLINICAL IMPLICATIONS

Although the study findings indicate that each participant has a unique story to tell, their HD experiences can be unified by the storylines of Quest, Chaos and Resignation. It is hoped that by identifying these HD storylines clinicians can be assisted in understanding the HD illness experience. Ricouer (1991) and Frank (2012) postulate that stories teach people who they are and allow them to know what to call their experience. If clinicians can learn to recognise how people with HD construct and interpret their illness experience, then a more patient focused approach in clinical management and treatment can perhaps be achieved.

THE VALUE OF LISTENING

This study might help inform the training needs of clinicians on how to deal with 'chaos' and other difficult stories. By listening to patients' stories, not only can we learn from their personal experience, but also help raise issues around power differentials in the wider community, assert the value of lay knowledge and go some way to realising the potential for the patient experience to inform better health care (Gregory, 2010). In the context of HD, the participants' stories reveal shortcomings in terms of how clinicians respond and deal with the concerns of people with HD and their families. Examples include how clinicians disclosed the HD diagnosis and how clinicians responded to participants who sought assistance for managing HD behavioural symptoms. In many instances, the participants felt the clinicians they dealt with 'didn't care' and they 'felt dismissed'. Chaos narratives are difficult to listen to (Frank, 1998a) and clinicians may not feel prepared for such work (Egnew, 2018). The barriers to optimal care include clinical environments that devalue illness narratives (Kleinman, 1988). Clinicians prefer to hear restitution stories (Frank, 1995), where there is an immediate resolution or a solution for a problem. However, as Kleinman (1988) stressed an 'empathic witnessing' of patients' and families' stories of illness (p.10) is central to clinical work. Lang et al (2000) suggested that training clinicians with active listening skills allows deeper understanding of the patient's reasons for seeking care and can improve patient outcomes. An important point, in terms of this study is that the participants valued how other care professionals communicated with them. They understood that 'there are no quick fixes', and that HD is a complex illness. What they need is for others to duly acknowledge their experiences. Some illness stories are told to clinicians in exchange for care, and as Frank (1998) observed, often 'in these stories there is nothing to fix, only a great deal to listen to' (p.210).

STORYLINES IN CLINICAL PRACTICE

The HD storylines identified in this study could enhance the capacity of clinicians to listen to stories, like Jen's Martin's or Daniel's. The proposed typology could give clinicians 'a sense of what to listen for' (Frank 2020 p. 2). This framework could be a useful 'listening template' for clinicians in considering the ways in which people with HD construct their illness experience. Frameworks facilitate thinking, for without a framework a patient story 'can be easily dismissed as a one-off occurrence, a sad tale that has little claim to the listener' (Frank 2020, p.3).

The Quest storyline emphasises the illness as a journey, and that discovery and learning takes place in order to gain new insight. By giving attention to a patient's quest, the clinician can support them on this journey with timely assistance or give useful information to create a more collaborative endeavour, such as when coming to terms with the HD diagnosis. The clinician should not lose sight of the Quest that is centred on the person with HD . When decision making is impaired due to capacity or cognitive issues, the involvement of 'other persons with HD' and their stories are useful conduits to improve clinical care. Ultimately, the Quest starts and ends with the best interest of the person with HD in mind. Correspondingly, clinicians can act as a witness to help the person with HD reclaim their autonomy and sense of self in their illness and in their lives.

The next storyline, Chaos is the anti-narrative of restitution. This is where clinicians are 'either unable to understand what is wrong or unable to treat it successfully' (Frank 1998, p.201). In this study, HD chaos stories were intertwined with stress that exacerbates HD symptoms, also relational dynamics that caused family relationships to fracture. Although disordered, with a sense that no one is in control (Frank, 1995), Chaos stories should not be denied by clinicians, for this only intensifies the suffering (Frank, 2002; Donnelly, 2021). A clinician can provide recognition of this situation, this itself has its therapeutic benefits (Charon, 2007). The person with HD in the grip of Chaos needs someone who will just listen, without attempting any change. The problem, as Frank (1998) pointed out is how to honour the telling of chaos, while leaving a possibility of change and accepting the reality. Over-hasty offers of help often blind clinicians to the real picture of the illness experience (Frank 1998). Specific examples in this study include clinicians who have misdiagnosed HD, mistaking it for depression, and clinicians who provided hurried solutions with negative consequences. The main implication of the chaos storyline is to invite clinicians to simply listen. For when the story is well heard, it becomes possible for HD issues to be talked about and reflected upon, and for shared solutions to be devised that keep chaos at bay. For Frank (2008, 2010) and Charon (2012), the prescriptive function of illness narratives can be built on the understanding that illness is a moment at which changes, and solutions, are always possible.

The Resignation storyline can assist clinicians recognise how people with HD construct and reinforce their relationship with their illness and acknowledge the presence of it in their lives. The study findings concur with recent research (Layton et al., 2021; Zarotti et al., 2022) which emphasizes the greater need for attending more closely to the psychological and social aspects of HD. Clinicians could also help support the person to live a fulfilling life with HD through regular care assessments and provide continuity of care in the long disease trajectory. Clinicians could also consider how resignation can provide new perspectives on present needs, and on the constant adjustments and changes brought about by the illness. More importantly, the clinician should also involve the spouses, caregivers and families and listen to their stories on the possible ways to recognise both quest and resignation, at the same time balance the needs of everyone affected by HD.

When people are encouraged to share their stories, it becomes possible to identify the strengths and coping strategies people have used, and which they may not realise they had. The participants in this study were able to make sense of their HD illness experience and identified their own capabilities and resilience to deal with the impact of HD on their lives. I suggest that clinicians should embrace the stories people tell about their illnesses to obtain a deeper understanding of the person behind their illness, and their reasons for seeking clinical care.

The use of narratives in clinical settings and research has been widely advocated in relation to chronic and critical illnesses (Charon, 2012; Molzahn et al., 2019; Ellingsen et al., 2021). More recently, narratives have been used in HD through genetic counselling sessions and group therapy to produce favourable outcomes in facilitating emotional support after receiving a positive (Stopford et al., 2020) or negative (MacLeod et al., 2018) predictive test result. Zarotti et al (2021) issue a call to arms for more psychological interventions for people with HD and follow this with a five-point manifesto which begins with 'Listening to people with HD' (Zarotti et al 2022, p.1.). I agree and further suggest that HD clinicians should commit and truly listen to the narratives of illness experience. In the end, what the patient brings to the clinician is their stories. As Egnew (2018) suggested, in chronic and fatal diseases where restoring health, relief of suffering or cure may not be always possible, the clinician's role is 'less than a problem-solver but an accompanier' (p.164). Like Frank (1995), Klienman (1998), Charon (2001), Sacks (2014) and Bird (2021) all remind us, patient stories not only reveal the content and meaning constructed around illness but help us enter a therapeutic relationship, to become a witness and provide better care for the person.

STRENGTHS OF THE STUDY

A major strength of the study was the use of narrative methodology, which allowed the participants to be active contributors to the study, this has given them a voice and added their stories to available knowledge. This permitted the person with HD and their family to fully express their experiences in a research setting. Moreover, to my knowledge, this is the first study to apply Frank's (1995, 2010) approach to exploring the illness experience of people with manifest HD symptoms. The study demonstrated the flexibility of the methodology to adapt to the needs of the participants, regardless of their HD stage.

Another strength of the study is that it was inclusive through using different communication aids. The multi-modal narrative methods catered to the HD physical, cognitive, and behavioural symptoms, and the study was also adapted to the mutable issues of research, such as changes with participants' communicative abilities and the conduct of research under Covid-19 restrictions. A specific example can be seen in the case of Jen Walters. Jen's speech deterioration posed a challenge to the traditional oral interview methods, and this was further highlighted with the pandemic restrictions where we had to resort to use voice over internet protocols (VoIP). It was challenging to fully capture Jen's non-verbal signs of communication in remote interviews, in comparison with face-to-face situations. However, when Jen used the narrative resources available to her it elicited meaningful data that made it possible for Jen to continue with the telling of her story. Initially, I also felt that Jen's physical and cognitive symptoms might hinder her

research contribution, but rather than dwell on her limitations as an informant, I focused on ways to address the barriers (Booth and Booth, 1996). Firstly, Jen's ability to recollect past experiences had diminished, but this lessening sense of self-continuity through oral discourses has been filled in by the accounts of people who care for her which consequently brought rich insight to her story. Secondly, Jen had to seek physical assistance during interviews, such as the holding her Ipad, which fell upon the care home staff, but this gave me the opportunity to observe aspects of care and how she interacted with her caregivers, and how this changed over time. Finally, the use of communication aids and personal artefacts was a useful intermediary in our interaction. What this study further adds to our knowledge of the practicalities of conducting research in HD advanced stages in cases such as Jen's, is to recognise their physical and cognitive impairments (Carmichael et al., 2019; Zarotti, Simpson and Fletcher, 2019) and use tailored methods to accommodate them. The study has combined relevant methods in learning disabilities (Booth and Booth, 1996), dementia (Phillipson and Hammond, 2018), and narrative studies (Buchholz, Ferm and Holmgren, 2018; Diehl and Riesthal, 2019), all which were useful to a story for people in late HD stages, such as Jen's.

The ethical rigour of the study can be viewed a strength. The study has undergone lengthy and detailed ethical approvals. This is due to the consent and capacity issues associated with HD symptoms (Wilson et al., 2010) and the sensitive domain of the research (McCosker et al., 2001). The study necessitated that any risk to the participants was minimised (Appendix G) albeit emotional risk was, to some extent, endemic to the research. However, the issues that arose during the conduct of the research as the Covid-19 pandemic took hold, were considerable and non-negotiable (Ferini-Strambi and Salsone, 2021, Cardel et al., 2020, Lebrasseur et al., 2021). Charmaz (2002) noted that, 'current challenges to the researcher's stories include the nature of our relationship with those we study, our representation of them and their stories, and the ethics of observing and reporting' (p.319). I addressed these ethical issues with openness about my own position in the research process, exercising reflexivity and transparency in my reporting and employing comprehensive risk assessments. Moreover, I outlined in my risk assessments (Appendix M) and undertook sufficient steps to provide a safe space for myself and the participants. I was able to continue with the study despite the pertinent issues of conducting research at the height of the pandemic.

CHALLENGES AND LIMITATIONS

The main challenge in undertaking this research was the recruitment and retaining of participants. Despite my professional affiliations working with HD patients, it was difficult to find participants suitable for the study. HD is a hard-to-reach population due to the rarity of the condition (Roos, 2010), and its many associated health issues (LaDonna and Ravenek, 2014). The careful selection of appropriate participants and the maintenance of a close link to patient support groups such as the HDA, gave me a measure of confidence that support was available to those who took part, should any issues arise in the course of the research.

The unforeseen challenge was conducting the study at the height of Covid-19 pandemic. The participants repeated cancellations of the interviews led me to question if they were still willing to engage with the study. Studies conducted in 2020 found the reduced research commitments of participants and their lack of interest in engaging with researchers (Cardel et al., 2020; Saberi, 2020; Vindrola-Padros et al., 2020). In a large mixed methods study of 250 participants involved in health research, Cardel et al (2020) found a link between the perceived threat of Covid-19 and the degradation of participants' mental health outcomes which has affected research activities. These observations were not surprising given my correspondence on the subject based on my contact with other doctoral researchers in various UK universities. These discussions arose in online research seminars concerning the impact of Covid-19 on qualitative researchers. Not surprisingly, a 2020 survey of UK PhD students found that the successive lockdowns have negatively impacted more than 89% of research data collection (Lambrechts and Smith, 2020). Despite the precariousness of interview schedules affected by Covid-19, the remote data collection continued with the use of life story templates and other documents and artefacts (Appendix D). This has also been made possible by maintaining communication either through phone or email and providing reassurance to the research participants. These challenges were overcome by keeping the lines of communication open, being flexible with dates and with frequents checks on the participants.

Another particular research challenge was the transition from face-to-face interviews to the use of remote technologies. This was not an easy transition for the HD participants, exemplified by Jen, who needed her caregiver to support her in using her iPad. Moreover, Martin and Daniel admitted they found it difficult to use some features of mobile technology. Martin preferred paper copies and did not use his email, and Daniel admitted he was 'finding it hard' to use 'Zoom and all that'. By contrast, internet and mobile technology facilitated interviews with HCPs and with Jen's children, Timothy and Marjorie. What this study highlighted were the disparities and increasing technological gap between people with HD and non-HD. However, providing options such as life story booklets and other personalised methods of data collection allowed the study to continue in the wake of pandemic restrictions.

In terms of limitations, the storylines described in this study do not encompass all features of the participants' illness experiences nor represent their overall HD story. Like Frank (1995), I acknowledged the limited application of the storylines, which should not be seen as an attempt to finalise or limit the capacities of the individual stories. The stories of Jen, Martin and Daniel should never be considered final, rather a contribution to 'an ongoing process of reassembling what will never be a whole story' (Frank 2010 p. 103).

Illness narratives are fluid and complex in nature. Storylines can be found alternatively and repeatedly at any juncture of the illness (Frank, 1998a). I take the view that what can only be described by the storyline was limited to what predominated at that moment or in this case, the HD participant's illness narratives at the time of the study. What was told in the participants' stories is unfinalized and 'acknowledged never to be the full story' (Frank 2019 p. 15). Frank likened storylines to 'patterns in a kaleidoscope, for a moment

the different colours are given one specific form, then the tube shifts and another form merges' (Frank 1995 p.76). Storylines are not fixed but rather reflect many levels of meaning. In this study, storylines were used as pathways to grasp the stories of people with HD. Staying with a story is akin to 'finding a usable pathway through a thicket' (Frank 1998 p.200), and as others leave these recognisable pathways, new ones are found.

RESEARCH EVALUATION

Creswell and Miller (2000) asserted that qualitative inquirers need to demonstrate that their studies are credible. Others have argued that this should be differentiated depending on the methodology used (Rolfe, 2006; Onwuegbuzie and Leech, 2007; Polkinghorne, 2007). Bocher (2000) suggested that 'multiplicity of goals implies multiplicity in standards of evaluation' (p.268). The debates continue and new criteria emerge amongst scholars on what determines the quality of a piece of qualitative research (Tobin and Begley, 2004; Noble and Smith, 2015; Carminati, 2018; Huttunen and Kakkori, 2020). Creswell (2007) provided clarity by offering specific criteria for narrative research (Table 14), and I will consider how they have been addressed in the study.

Table 14. **Narrative research evaluation criteria (adopted from Creswell 2007)**

Narrative research evaluation criteria (Creswell 2007, p. 214)	**Study evidence: Chapters and Sections of the thesis where criteria were applied**
• **Focuses on one, two or three individuals**	Chapter 5: Findings of the HD stories The illness experience of Jen, Martin, and Daniel
• **Collects stories about a significant issue related to the individual's life**	Chapter 3 and 4: Methodology and Methods. The use of BNIM, life story template, artefacts, and documents.
• **Develops a chronology that connects different phases or aspects of the story**	Chapter 4: Methods and Narrative Analysis Chapter 5: Presentation of narrative findings
• **Tells a story that re-stories the story of the participant in the study**	Chapter 4: Methodology and Narrative Analysis Chapter 5: Narrative Findings Appendix L (story images, letters, appreciation of the narratives)

• **Tells a persuasive story told in a literary way**	Chapter 5: Findings: HD stories
• **Possibly reports a theme that builds from the story to tell a broader analysis**	Chapter 5: Findings: HD stories structured in storylines Chapter 6: Discussion relating to the study findings and literature
• **Reflexively brings himself or herself into the study**	Chapter 1: Researcher context Chapter 3: Section on epistemology Chapter 5: Findings, Introduction and Epilogue Chapter 6 and 7: Discussion, revisiting reflexivity and conclusion

In addition to Creswell's (2007) criteria, I believe the study was enhanced by extending the main finding of my MSc research that the 'HD patient is the biggest teacher' (Carreon, Hayes and Leavey, 2018). The methodological rigour of this study was enhanced by its longitudinal design, use of multiple sources of data and employment of narrative co-construction (Rolfe, 2006; Noble and Smith, 2015). This was facilitated through collaboration with the participants from the conception of the research design (Participatory Research Exercise) to the interpretation of the stories by sharing both the transcripts and the core stories.

Throughout the study, I have committed to the methodological, reflexive, and ethical stance which I have discussed in previous chapters. During the conduct of the research for this study I maintained a close link with HD stakeholders, including patient support groups, HD clinical groups, and advice was sought from the HDA advisors during the progress of the study to ensure the relevance and significance of the study to the HD community. This study has demonstrated the robustness of naturalistic inquiry with the use of the above pluralistic approaches (Lincoln, 1995; Tobin and Begley, 2004; Creswell and Poth, 2016). Most importantly, the active involvement of the participants not only highlighted the importance of sustaining rapport and negotiating meaning in the entire research process but also enhanced the trustworthiness (Birt et al., 2016) and validity of the study (Creswell and Miller, 2000).

Bruner (1989) argued that, a good story and a well-formed argument are different natural kinds. This claim was elaborated by Rubin (2014) to the effect that whereas good arguments convince you of truth, a good story endows experience with meaning, and I approached the analysis of the participants' stories by turning 'research data into a meaningful story' (Ellis 1995 p.313). Whereas Ellis (2000) provided insight into evaluating stories within ethnography. She admitted a chaotic process of questioning, thinking, and feeling with the stories until the material settled. Frank (2019) maintains that 'no clear guidelines can be laid down because the nature of stories is to elicit multiple interpretative reactions' (p.14).

Polkinghorne (2007) asserts that the storied descriptions and the meaning participants attribute to their life events is the 'best evidence available to researchers' (p.79). In the end, I assessed the participants stories with a Nietzschean interest beyond good and bad, but remained faithful to the participants' voices, I wrote with them in mind on how their experience and lives truly depend on.

SUMMARY

In this chapter I have discussed how the participants constructed their illness experience and applied Frank's (1998) Quest and Chaos storylines, with the addition of the novel storyline of Resignation. It has been suggested that the narrative approaches to and identification of these storylines can provide general and specific clinical implications to guide HD care. In the next chapter, I will conclude the study with reflexive accounts of the relational and ethical issues raised, a discussion of the contribution made to the literature, recommendations, and suggestions for future directions.

CHAPTER 7: CONCLUSION

For over four years, I have fully immersed myself in HD stories as my full-time job. During this period, I have come to better understand what Arthur Frank (2010) meant when he wrote, 'that stories are not materials to be analysed, rather a relationship to be entered' (p.200). In this final chapter, I will reflect upon these research relationships as I draw the thesis to a close. I will re-visit reflexivity and ethics, and I will examine key issues that have arisen during the research process to further contextualise the study findings. Finally, I make the case for the contribution that this study makes to the literature, suggest future directions, and offer my closing thoughts.

RE-VISITING REFLEXIVITY

Reflexivity provided insight into 'how knowledge is produced' (Pillow 2003 p.178) and became my personal lens to interrogate the intersubjective dynamics between myself and the participants. Here, I turn a critical gaze towards myself, my self-location (gender, race, class, sexuality, ethnicity), position and interests (PhD student, novice researcher, physiotherapist) (Berger, 2015) and make an assessment of how this has influenced all stages of the research process (Finlay and Gough, 2008), including my interpretation and representation of the participants' lives, through their stories (Etherington, 2007; Finlay, 2017). Like Finlay (2003), reflexivity became 'my solution and my problem' (p.3). It was my solution in the sense that it helped me make important study decisions, particularly in the methodology and analysis, but also my problem in that I had to make sense of my interpretation of the data and expose the dilemmas of my roles as a health professional and researcher.

In Chapter 1, I outlined my interest in the topic and assumptions to identify the subjectivities that could have influenced the research direction. In Chapter 4, I outlined my reflexive approach to the study design and during the pre-research stage, and how I conducted the interviews and collected data. My theoretical and epistemological position has guided the methods of inquiry and the analysis of the data. I am also aware that my personal and professional identities have had a considerable impact on the participants, data analysis, and outcome of the study. Illness narratives place significance on context (Murray, 2008; Riessman, 2008; Thomas, 2010; Kokanović and Flore, 2017), and while the study asked how people with HD construct their illness experience, in this section I will further elucidate the context of how the story is constructed (Josselson, 2011). Mishler (2004) argued that 'different researchers may tell different stories about what they claim are the same events in people's lives' (p.101). Josselson (2011) asserts that by practising reflexivity, the study places context to the 'findings as relative to the researcher's standpoint' (p.226). My standpoint in this study is articulated by Frank (2000) as an 'act of self-reflection' (p.356) on the relational and ethical decisions.

RELATIONSHIP TO THE STUDY: OUTSIDER-INSIDER STANCE

Frank (2010) issues a call to 'bear witness' (p.142) to people's illness narratives. However, to bear witness entails an invitation to enter people's personal lives. The challenge is how to be invited into a closed group

such as the HD community, where people's experiences of stigma, discrimination, and isolation might make them reluctant to invite an outsider in (Wexler, 2010; Williams et al., 2010; Halpin, 2018; Wauters and Van Hoyweghen, 2021). Maxted et al (2014) found that the HD family serves as 'a unit that protects those within but isolates those outside its borders' (p.346). On the face of it, I was an outsider; a single, gay, southeast Asian man exploring a predominantly Caucasian familial disease (Pringsheim et al., 2012; Bruzelius et al., 2019). Genetic neurological conditions do not run in my family; therefore, I have no direct experience of how families live with hereditary risks. I have no underlying health issues, and I cannot personally relate my everyday experiences to those of people living with chronic illnesses. However, I was also an insider in my professional identity as a specialist HD physiotherapist, with over a decade of work with neurogenerative diseases. The study was promoted through an HDA charity, in which I had established professional relationships with HD advisors. The charity provided two platforms for potential recruitment, a newsletter, and a support group. Although from a personal view I was an outsider, I was no stranger to the HD community as I was professionally connected with HD networks. This may have contributed to how the participants engaged and gave me access to their personal lives. I viewed my relationship to the study as a witness to the HD illness experience. As Dwyer and Buckle (2009) and Paechter (2013) have argued, it is not the outsider-insider stance of the researcher that matters, but their determination to keep the participants at the centre of the research, and their commitment to act throughout with integrity, authenticity, and good faith.

RELATIONSHIP TO PARTICIPANTS

I had established a good rapport with the participants, but I am aware that my relationships with them varied considerably. The married couple, Martin and Pamela, initially referred to me as 'still young with so many opportunities in life'. I was also mindful that every time I visited their home, they laid out sandwiches and cakes, and made sure that my return journey was easy, by offering money to pay for my bus fare. At one point Martin summed this up by saying, 'You're like family to us, Paul.' The relationship started from familiarity and moved to a place of trust, as Pamela said, 'I wouldn't tell anyone about this, but if this helps with your research.' Daniel, on the contrary, displayed a more casual approach. He was easy going and preferred to move our interviews to a cafe. Jen had previously been my patient and her family had known me from my clinical practice in the nursing home. Our interactions had moved beyond the domains of physiotherapy to that of their private life. One of my supervisors asked me, 'How do you feel about your relationships with the participants?' and I reflected on this question, that I actually didn't really think much about it. Rather, I was open and authentic in my approach and that guided how rapport was established throughout the study.

However, I was conscious of my feelings of appreciation for the participants. I constantly expressed my gratitude given the difficulty of study recruitment, and retention. Indeed, I was grateful to them for granting me access to their stories and personal lives. In the beginning, I viewed the participants as a trove of data waiting to be uncovered. But I then realised, people, like stories are not materials to be analysed (Frank, 2002), but relationships to be nurtured (Frank, 1998a). Peoples' illness experiences were more than

the words they provided, and much more than their disease and their illness narratives. Participants in research like this, who are offering up the fruits of their illness experience, need to be treated as autonomous individuals who come with their histories and identities. What is important is how to form a meaningful research relationship that is based on honesty in order to establish mutual trust. This was the basis of my decision to refer to the participants in this study by their names, as 'people' and not just 'data', recognising personal meaning and offering respect to their illness experiences. Conducting this narrative study taught me how to honour and value research participants and relationships, treating 'people as persons and not as objects' (Morse and Johnson, 1991 p.2), recognising their suffering and triumphs; and the realities of everyday living and of reconciling with a fatal illness.

SENSITIVE TOPICS

In the remote interviews, there were instances of sadness and distress that were apparent. Such are not uncommon in cases of research into sensitive topics (Dickson-Swift et al., 2007) and other researchers have written extensively about these experiences (Gair, 2002; Etherington, 2007; Hydén, 2008; Berger, 2015; Soilemezi and Linceviciute, 2018). In this study, a couple of participants expressed sadness and lapsed into an unexpected silence after discussing and revisiting difficult life events. On one occasion one of the participants became very emotional as I could hear her crying at the other end of the line. However, it is worth noting that none of the participants terminated the interviews, in fact most of the remote interviews lasted over an hour and all participants expressed their gratitude at the end of each interaction.

While researchers have noted the potential of sensitive topics to cause traumatisation (Elmir et al., 2011) and psychological harm (Mealer and Jones, 2014) that can significantly affect both participants and researchers (McCosker, Barnard and Gerber, 2001; Mallon and Elliott, 2019), Corbin and Morse (2003) argued that this level of distress is short lived and not greater than in everyday life. However, the existing literature on interviews on sensitive topics was produced pre-pandemic (McCosker, Barnard and Gerber, 2001; Corbin and Morse, 2003; Dickson-Swift et al., 2007; Mealer and Jones, 2014; Friesen et al., 2017; Mallon and Elliott, 2019). Despite the fact that Covid-19 ethical guidelines were produced, they chiefly focused on clinical trials (Dawson et al., 2020; Saberi, 2020; Jamrozik et al., 2021) and remote online data collection (Chatha and Bretz, 2020; Vindrola-Padros et al., 2020). In the era of socially-distant methods (Lobe, Morgan and Hoffman, 2020), more research is needed on the use of qualitative inquiry in the cases of specifically vulnerable participants with serious health conditions.

The impact of Covid-19 pandemic caused serious medical and social disruption (Teti, Schatz and Liebenberg, 2020). The increasing literature has established the psychological and physical impact of Covid-19 restrictions (Brooks et al., 2020; Cardel et al., 2020), which disproportionality affected people with disabilities (Turk and McDermott, 2020), people with mental health and neurological conditions (Mesa Vieira et al., 2020; Ferini-Strambi and Salsone, 2021) and people in long-term care (Velayudhan, Aarsland and Ballard, 2020). This led me to consciously adopt a deeper ethical attitude (Josselson, 2007), trying to become 'more humane' (Mishler 2005, p. 437) and 'being present' (De Marrais 2003, p. 67) in my inquiry.

My position remains that of a health professional and a researcher, with a personal commitment to maintain sensitivity and balance in my attention to the realities of the situation (Corbin and Morse, 2003; Charon, 2012), to the well-being of the research participants (Social Research Association, 2021) and to the wider societal consequences of the pandemic (Mesa Vieira et al., 2020; Teti, Schatz and Liebenberg, 2020). Carlson (2020) places particular significance on the practice of relational ethics at the time of Covid-19, highlighting that those who conduct a narrative inquiry must have the 'courage and compassion to pursue other ways of knowing and being in the world' (p. 1149).

Despite the recent update of the ethical guidelines for social researchers (Social Research Association, 2021) there is a lack of literature on conducting sensitive topic interviews at a time of health crisis such as the Covid-19 pandemic. I remain guided by Corbin and Morse (2003) who assert that when the research is committed to ethics and conducted with sensitivity it will benefit both the participants and the researchers. Denzin (2014) reminds us of the responsibility that arises from the conduct of this kind of research; to the people and the communities we are studying. At the end of each interview, I confirmed with participants if they had access to appropriate social or professional support including details of the research signposting services (Appendix I). Santana et al (2021) and Friesen et al (2019) advocate that researchers observe ethical values in their research activities. In the end, my worry about causing anxiety and distress to the participants did not materialise, even in the cases of the two interviews that had to be paused. One participant, Daniel said, 'I've enjoyed that. It's been good' and Marjorie, a family member, concluded her interview, 'a humbling experience, thank you for listening to me'.

IMPACTS ON THE RESEARCHER

Most of my data analysis was conducted at the time of working from home and social restrictions were being implemented. Aside from trying to understand how the rest of the world was making sense of the pandemic, my main task of the day was to work out how to make sense of the illness stories I had heard. Often, I felt too close to the data, conscious of a mainly emotional response and of feeling drained after I read the transcripts and listened to the recordings. Although common in researchers studying sensitive topics (Dickson-Swift et al., 2007), my feeling of paralysis when contemplating the analytical task was augmented by the variety and amount of data I had collected. Kramp (2004) warned that engaging in narrative research calls forth the researcher's 'most exacting abilities', for it is 'laborious and time-consuming' (p.113). I was cognisant that a narrative approach requires rigorous collection, collation and synthesis of data followed by critical analysis, reflection, and reflexivity. For Bleakely (2005), narrative research demands a 'high level of ethical and critical engagement' with participants and the data (p.539). However, I hadn't accounted for the pandemic, where suffering, death, and illnesses were the dominant discourse of everyday life. At the time, I felt I had no relief from these topics.

Clandinin and Murphy (2007) drew attention to the 'loneliness of the work of narrative inquirers' (p.7) and suggested the importance of sharing work with other researchers. This was perhaps one of the reasons why I delivered several virtual presentations at this time (page ii). Looking back, several factors could have

contributed to my personal challenges with the study data. These could include the sensitive topic around illness experience, narrative methodology, and being a PhD student at the time of the pandemic. However, what helped me to stay focus was the use of the elaborated method of analysis and the questioning guides (Figure 22, Chapter 4), this dialogical approach to narrative analysis (Frank, 2002; Riessman, 2008; Frank, 2010) kept me on track on how to navigate the above issues. I was able to distance myself from the study, evaluate my relationships with the participants and adopt multiple perspectives to generate findings with greater clarity.

The narrative study developed what Josselon (2007) referred to as an 'inherent relational endeavour' (p. 537). Most interviews were conducted at the time of the pandemic, where physical and social restrictions were imposed by the government. For the participants, I became one of their regular social contacts. Campbell (2021) reflected on how his narrative study had created an 'unexpected intimacy' with his participants, where the researcher spoke of 'joyous occasions' and interactions that involved participants and their children around family life (p.576). This could not contrast more strongly with my experience during this period, in which most of our research conversations were deeply emotional, and often I found myself worrying about my participants' personal problems, vulnerabilities with health issues and the risk of Covid-19. I was torn between my role as a researcher and that of a health professional in that I often felt the need to protect the participants or do something about their health issues. Nonetheless, I was guided by the participants on how they wanted to continue with their participation, and they reassured me that they were happy for the interviews to resume. Consequently, I ensured that at the end of each interview the participant had access to signposted support or was directed to their GP if needed.

Ethics in narrative research, as Josselson (2007) points out, 'is not a matter of abstractly correct behaviour but of responsibility in human relationships' (p. 538). I maintained a researcher stance with a 'compassionate distance' (Gabriel et al. 2017 p.163), engaging in a research dialogue of 'becoming close without merging' (Frank 2009, p. 108). The supervisory discussions and my reflective journal had helped me make sense of my researcher role and of my relationships with the participants. By these means I was able to establish the importance of timely supervisions to address these relational issues and to maintain self-reflexivity. The research process was not neat and linear, but my adoption of Gabriel's (2009) guidelines reassured me that I had taken the necessary steps to support the participants and that the risk of research role-conflict was minimised (Table 15).

Table 15. Guidelines to minimise researcher-role conflict (adopted from Gabriel 2009)

Guidelines for minimising researcher-role conflict	Requirement met via (where applicable):
1. Provide clear information for participants about: • Details of the research • Informed consent • Information on the possible consequences of participating on them, the relationship and maybe others too	✓ Research study website: https://voicesofhuntingtons.wordpress.com/ ✓ Informed consent forms ✓ Study information forms ✓ Participant forms ✓ Pre-interview discussions
2. Form an effective research alliance by: • Negotiating a clear contract with good boundaries • Keeping a compassionate distance in the research role • Outlining the tasks and goals of participant and researcher • Communicating limits to the relationship (e.g., what happens when the research is over?	✓ Informed consent forms ✓ LJMU research ethics codes of conduct ✓ Study information forms ✓ Pre-interview discussions ✓ Participant forms ✓ Pre-interview discussions ✓ Exit interview discussions
3. Have a clear policy on confidentiality: • Point out the context of what is disclosed in the research interview is not subject to counsellor/supervisor codes of ethics • Be clear about the limits of confidentiality and when you might have to discuss the information with others	✓ Study information forms ✓ Informed Consent forms ✓ LJMU ethics policy framework ✓ LJMU research ethics codes of conduct ✓ Interview discussions
4. Cultivate self-reflexivity by: • Regularly reviewing the researcher role/research process • Using code of ethics for researchers and moral principles etc. for guidelines for consultation	✓ Supervisory meetings ✓ Researcher journal ✓ Interview discussions ✓ LJMU ethics codes of conduct ✓ LJMU ethics policy framework ✓ UKRI ESRC research ethics guidance

MY POSITION ON SILENCE AND UNSPOKEN NARRATIVES

The paradox of this study is that it revealed (and concealed) other narratives, meanings, and interpretations. It is my view that a narrative ends with some parts missing, for it is composed and interpreted, rather than remembered. I also respected the unspoken narratives that participants chose to keep for themselves. These include topics around previous relationships and around genetic status. Additionally, there were stories that were omitted and not included in the writing up of this thesis due to its highly sensitive and identifiable content. I took full responsibility for what is written up, and I respect what was never said, for silences provide their meaning too. For as long as I conduct ethical research, this understanding will give me guidance on 'which questions to ask, which secrets to keep, and which truths are worth telling' (Ellis 2007a, p.26).

The silences refer to Mazzei's (2003) quest to discover the truth in stories, where there are things intentionally repressed. Eliciting the stories for this study came with silences or unspoken narratives, and rather than viewed as data gaps or as the interviewer's failure (Mazzei, 2003), I understood my participants' silences as values (Poland and Pederson, 1998) that needed to be discerned (Charmaz, 2008). In my interactions, the participants' silences invoked me to be attentive to what goes unsaid (Booth and Booth, 1996; Poland and Pederson, 1998) and listen to what is less directly expressed (Mazzei, 2003). For Charmaz (2008), silence has implicit meanings, and we must 'learn what they mean and where they reside' (p. 16). There were silences around the previous relationships of Jen and Martin, also concerning Daniel's lack of intimate relationships. In comparing HD with other neurological illnesses such as PD and MND, researchers have found that HD has significantly lower intimate relationship satisfaction (O'Connor, McCabe and Firth, 2008). Silences also hover around the genetic status of Timothy and Marjorie, the question of whether they have been tested, meaning that secrecy around their HD status persists. These silences were purposeful and meaningful (Mazzei, 2003) so I left them as such. In their silence, 'there is nothing to fix, only a great deal to listen to' (Frank, 1998, p. 210). Rather than dwell on the absence of words in these intimate and confidential moments, we should 'grasp the larger story, and we may need to be silent' (Charmaz 2002 p.323) for all of it reveals the fullness of a story.

WITHDRAWAL FROM THE FIELD

I attempt to balance my relationship and involvement with the participants and my responsibility to uphold standards of academic scholarship (Josselson, 2007; Gabriel, 2009). In the above sections, I discussed my ethical approach to interviewing and each time I completed an interview, I was impelled by my desire to end on a positive note. A wealth of interview studies has documented the therapeutic effect and catharsis that participants feel when they share their illness experiences (Charmaz, 1991; Charon, 2001; Dickson-Swift et al., 2007; Ellingsen et al., 2021). In this study, the participants showed their appreciation of being involved in the research. I was confident that I abided by the ethical rules of conduct, of informed consent, confidentiality and privacy, and have protected participants from harm. However, as Ellis (2007) and Josselson (2007) argued, the actual research dilemmas of ethical attitude (Josselson, 2007) or relational

ethics, related to ethics of care (Ellis, 2007) are seldom talked about and are not the focus of institutional ethics applications. In this study, the practical and relational issues were centred on the research practice and the changing relationships with the participants over time. I sought guidance from ethical guidelines (Social Research Association, 2021) and from the experiences of other researchers (Frank, 1995; Mishler, 2005; Dickson-Swift et al., 2007; Adams, 2008; Damianakis and Woodford, 2012; Bruce et al., 2016), but just like Ellis (2007) I have 'struggled with ethical choices time after time' (p.5).

The question I returned to as I drew the study to an end, was 'If our participants become our friends, what are our ethical responsibilities toward them?' (Ellis, 2007, p.5). This ethical dilemma is innate to narrative research (Clandinin, 2006; Josselson, 2007), as it is to studies that involve work with vulnerable people, that focus on sensitive topics, or those that involve prolonged contact, such as multiple interviews over time (Dickson-Swift et al., 2007; Etherington, 2007; Andersson et al., 2016). When I conducted my exit interviews, all participants wished me luck with the study and the rest of my career. However, in my attempt to withdraw from the field, I realised that while I might leave the field of research, I couldn't sever the human field of connection and relationship. Martin and Pamela said, 'let us know how you get on', Daniel's words were 'keep in touch mate', Timothy remarked, 'if there's anything else you need, let me know' and Jen ended with 'see you soon'. Josselson (2007) warned that researchers must be prepared for the fact that participants might wish to continue with the relationship, and that there is 'nothing unethical if this wish is mutual' (p.545). I will remain in touch with the participants if they continue to wish it. However, I will gradually limit our conversations around research related topics to wean them off the researched-researcher relationship. By engaging in this type of research, I was able to gain a greater understanding of my ethical decisions, and the kind of researcher I want to be. The research relationships that form the basis of this study had started when I stood as a witness and entered a relationship with Jen, Martin, and Daniel. They were unfinalized storytellers living with a terminal illness, and their narratives of HD illness experiences have opened a dialogue of how listeners can take part in 'the concentric circles of witnessing suffering' (Frank 2013 p. 195). If they wish for me to witness the rest of their illness journey, I will be that person who Frank (2010) advocates to stand beside them.

CONTRIBUTION TO LITERATURE

This study has filled a gap in the existing HD qualitative literature, specifically by exploring HD experiences beyond period of diagnosis and onset of symptoms (Hagberg, Bui and Winnberg, 2011; Audulv, Packer and Versnel, 2014). It has given an insight into the psychosocial aspect of the illness (Halpin, 2018; Mahmood, Law and Bombard, 2021) and has deepened our understanding of how HD impacts everyday life (Audulv, Packer and Versnel, 2014). Although limited to a small sample, the study responded to the call for 'more qualitative studies exploring subjective experience of persons with HD' (Zarotti et al. 2022, p.5), to employ multi-methods (Audulv, Packer and Versnel, 2014), and for a longitudinal approach that could give an insight into understanding people's life decisions and illness perceptions (Helder et al., 2002; Quaid et al., 2010). The use of tailored methods addressed the participants' changing needs, as well as Covid-19

restrictions. Moreover, by adopting a narrative approach hitherto not applied to HD but widespread in the study of other chronic illnesses, I have brought a novel perspective to better understand the HD illness experience.

Secondly, the study has deepened our understanding and knowledge of the complexities of living with HD. At a more intimate level, the study explored the personal impact of HD on the person, their relationships, and their families. HD needs to be viewed as more than its genetics or clinical presentation and understood in the context of how the illness is experienced by the person, especially in their relational and psycho-social aspects. The study illustrates the variety of ways HD narratives can be a useful resource in providing context and perspective to the person's predicament, particularly for clinicians who wish to provide better care. Whilst the study provided a detailed contextual narrative analysis on HD at different stages, it also highlighted the common unmet care needs of individuals and their families.

This study supports the need for tailored care planning, which would include both service provision all throughout the disease trajectory, and the acknowledgment of the complex changing disease feature. Beyond addressing the HD pathophysiological needs, psychological services and emotional support such as counselling should also be offered to affected individuals, including HD spouses and children. HD as a genetic illness makes demands of and affects the whole family system (Rolland and Williams, 2005). The study supports systematic viewpoints published in the last twenty years that have clearly established that HD care interventions should include families and caregivers (Brouwer-DudokdeWit et al., 2002; Aubeeluck and Moskowitz, 2008; Huniche, 2009; Jona et al., 2017; Parekh, Praetorius and Nordberg, 2017; Martins et al., 2018).

Finally, the participants' narratives have provided heuristic materials that I plan to develop into publications that will contribute to an area largely under-investigated in literature, which is on HD in the advanced stages. The examination of participants' narratives in this thesis addresses the broader HD knowledge gap in understanding HD subjective experiences (Zarotti et al., 2022) as 'described in their own voices' (Mahmood et al 2021 p. 15). By privileging the voices of Jen, Martin, and Daniel, this study opens up a dialogue with other HD research stories and their representations. I believe this study contributes to an 'ongoing process of reassembling' (Frank, 2010 p.103) the stories of HD illness experience.

RECOMMENDATIONS AND FUTURE DIRECTIONS

This study has contributed to our understanding of how people live with HD and how they narratively construct their illness experience, however there are still areas that require further research.

- The illness storylines of Quest and Chaos, with the novel addition of Resignation, were identified in the stories of the HD participants. I recommend that this narrative concept be applied to a variety of HD participants, considering specificity with age, gender, and sex differences due to their impact on disease symptoms, severity, and progression. Specifically, further exploration is

needed of the experiences of female HD sufferers to better understand the heterogenous nature of their illness experience. This is due to the more rapid disease progression in females, their poorer functional outcomes and their frequent adoption of heavier family and caregiving responsibilities (Martins et al., 2018; Zielonka et al., 2018; Ullah et al., 2019). In addition, the conduct of narrative research that addresses the full range of sociodemographic characteristics including ethnicity, co-morbidities, and economic status would create a more diverse data set and extend our understanding of the illness experience and care needs. This will give voice to and give us the opportunity to hear the unheard stories of the wider HD population.

- I suggest the development and delivery of training programmes for clinicians that focus on listening both actively (Lang, Floyd and Beine, 2000) and empathically (Kleinman, 1988). HCPs need to value narratives in clinical settings and identify strategies on how narratives could be integrated into the delivery of person-centred care, as Frank (1998) framed it, 'to give the gift of listening is the beginning of clinical work' (p. 200). This study was not envisaged as a care intervention, yet the narrative approach provided a therapeutic process within which participants could reflect on their illness experience and it would appear to have provided them with a positive experience. Personal stories and reflections could be used in clinical practice and other care contexts (Charon, 2012), and this would allow increased engagement and dialogic praxis for care professionals to better address the respective personal concerns of people with HD and their families.

  Further exploration on how HD services are organised, and how HD care is delivered is a research necessity. There were wide disparities in participants' experiences with various HCPs and health and social care institutions that this study has highlighted as unmet needs. This can be attributed to the scarcity of specialist HD services, the poor understanding of the disease amongst care professionals, and the lack of effective medications to delay the disease progression (Nance, 2007; Novak and Tabrizi, 2011; Frich et al., 2016). The detection and treatment of HD symptoms is, necessarily, a matter for both psychiatric and neurological services (Halpin, 2011; van Duijn, 2017) but this study has noted that they are often not well coordinated. Studies in the UK (Rae, McCann and Miedzybrodzka, 2014; Wilson, Pollock and Aubeeluck, 2014) and internationally (Etchegary, 2011; Frich et al., 2016; van Walsem et al., 2017) reported that HD needs were not adequately met by existing health care and social services. The experiences of Jen, Martin and Daniel reflect that the person with HD can easily fall between the gaps in care provisions. Despite the wealth of literature and guidelines on disease management (Mestre and Ferreira, 2012; Frank, 2014; Bates et al., 2015; Moskowitz and Rao, 2017; Patrick and Ritchie, 2020), including improvements in HD standards of care (Simpson and Rae, 2012), better understanding of the importance of MDT care (Nance, 2007; Frich et al., 2016; Edmondson and Goodman, 2017) and the availability of recommendations on the role an HD specialist nurse or key worker should play (Baker, McLaren and Crichton, 2009; Kenny and Wilson, 2012; Wilson and Aubeeluck, 2016), these interventions have not directly translated into the experiences of the study participants. More research is

needed to understand how people with HD interact with care services and how this affects their illness experience.

- Health services for people with neurological conditions were severely disrupted during the Covid-19 pandemic (García-Azorín et al., 2021), leading to further disparities of care and the exclusion of people with health-related vulnerabilities (Velayudhan, Aarsland and Ballard, 2020; Lebrasseur et al., 2021; Pfalzer et al., 2021). Remote assisted technologies such as telemedicine were suggested to mitigate the effect of the pandemic on neurological care (García-Azorín et al., 2021), but as illustrated by the participants' experiences and findings from an evaluation study in the United States (Pfalzer et al., 2021), a 'significant portion were not willing to participate' (p.313) in remote HD health services. Jen, Martin, and Daniel's health appointments were repeatedly cancelled because of the Covid-19 restrictions, and the way health services had to be delivered. At the time of writing, they have not received dates for their next HD assessment reviews. A new direction in research is urgently needed on the broader effect of the national pandemic restrictions and institutional responses to them. Insight into the experiences of living with PLTNC such as MS (Moss et al., 2020; Chiaravalloti et al., 2021), PD (Simpson et al., 2021) and people with dementia living in care homes in England (Tuijt et al., 2021) during the pandemic has been published, and in line with this momentum, further research on HD in the context of the pandemic is needed. Understanding the relationship between neurological conditions, the pandemic and the consequences for the general health and wellbeing of patients will help in the planning and preparedness for future pandemics.

FUTURE DISSEMINATION AND IMPACT ACTIVITIES

I plan to disseminate widely the study findings and highlight the importance of communication, listening and recognising each person's uniqueness and complexity through their narratives. So far, this work has been shared with physiotherapists and HCPs in conferences, educational sessions, and webinars of national and international HD organisations. This was made possible through my liaison with the HD Physiotherapy networks, HDA England and Wales and my role as a Workstream Lead of the European Huntington's Disease Network (EHDN) Physiotherapy Working Group (PWG) in Residential Care.

Extending this knowledge gained from the PhD study and beyond physiotherapists working with people with HD, the study insights were also disseminated at a Professional Certificated Training course for HD held in summer of 2022. The certificated professional course was organised by the HDA England and Wales where I was one of the keynote speakers who delivered the training course. Together with an HD neurologist, genetic counsellor, clinical psychologist, and other HD professionals, I provided a session on the person-centred approach to physiotherapy in people with HD and their families.

Furthermore, the understanding gained from the study, particularly on the use of narrative case studies, will be used to develop resources and training materials pertinent to physiotherapists working with people with HD advanced stages in long term care. In September 2022, I presented my research findings to the

EHDN PWG and I will continue to work with them to ensure my recommendations inform the physiotherapy practice. As a result, a series of educational sessions are planned until the end of 2023 to promote awareness of physiotherapy guidelines and best practice in physiotherapy in HD. In my role as Workstream Lead of EHDN PWG in Residential Care I will ensure that my findings inform the production of the HD educational materials. These materials will promote physiotherapy person-centred care planning, resonating with the adapted illustration which was inspired by the PhD conceptualised study case design (Figure 14, page 73).

Through working with HD charities, professional organisations, and networks of specialist HD physiotherapists I will promote better awareness around the disease and provide better clinical care for people with HD and their families. Moreover, I will continue to advocate for people with HD and produce peer reviewed papers derived from this PhD study to contribute to an ongoing dialogue of HD stories and how to use these stories of illness experience to help restore the person at the centre of their care. I am planning to write several papers originating from this PhD targeting key publications such as the Journal of Huntington's Disease and the Physiotherapy Journal published by the Chartered Society of Physiotherapy (CSP). In addition, I am very keen to publish a methodology paper likely to be submitted to the International Journal of Qualitative methods (SAGE) on the novel narrative multi-methods used in this study. I will also work with the editors of Frontline (also published by CSP) to write a summary paper containing key practice-relevant recommendations.

Physiotherapy practice is currently dominated by a biomedical perspective which can limit the adoption of person-centred care (Mudge, Stretton and Kayes, 2014). Mari (2019) observed that from a physiotherapy perspective, there is a lack evidence-based gaps on how physiotherapists integrate person centredness in the practice. In a study with older people with dementia, Hall el al (2018) found a lack of person-centred approach and shared understanding of the role of physiotherapy. Moreover, a systematic review of physiotherapy in musculoskeletal conditions, Naylor et al (2022) recommended that a more narrative approach to physiotherapy assessment can help maximise person-centredness. There is evidence to suggest that physiotherapists need to better engage in the practice of delivering a person-centred approach (Mudge, Stretton and Kayes, 2014; Mari, 2019; Naylor, Killingback and Green, 2022). This method of care is not intended to replace our clinical work, but to complement and balance our physiotherapy practice. It is my view that knowing the person's story and working with the person are vital means to contribute to the delivery of a person-centred approach to clinical care. Person-centredness in physiotherapy involves more than assessments and treatments, and clinicians might need to, metaphorically, stand beside the person to witness their illness journey and acknowledge their illness storylines. As the prominent neurologist Oliver Sacks (2014) said, we must work to deepen our clinical case notes to a 'narrative or a tale; only then do we have a 'who' as well as a 'what', a real person in relation to the disease - in relation to the physical' (p.1).

CLOSING THOUGHTS

Completing this study allowed me to reflect on the lessons that I have personally learned from the stories of the participants. I considered my previous assumption: that as a clinician, my main task was to rehabilitate and optimise the physical function of the body. My understanding from the literature (Bates et al., 2015; Ghosh and Tabrizi, 2018) was that a diminished quality of life in the case of HD was due to a failing body and mind (Huntington, 1872). I hung on to a Restitution narrative (Frank, 1995) for my HD patients, holding out the hope that treatments could restore, if not help them stay well. I clung to the 'single-minded telos of cure' (Frank 1995 p.83) and the heroic narratives of medicine and technology. However, in the case of HD, and this is perhaps transferrable to other life limiting diseases, there is no reprieve for the illness's inevitable deterioration. Throughout this study when I closely listened to the storylines of Resignation, I found the way the people with HD constructed meaning, accepted, and claimed their illness experience. This generated a new personal understanding that despite living with an unremitting illness and in the absence of cure, people with advanced HD can still live happily and have some degree of quality in their life. This is possible if they are understood as a person and not their disease, their personal stories are listened to, and their illness experiences are acknowledged. Equally, if their caregivers and families are appropriately supported, this will enhance their social needs, environment, and care delivery.

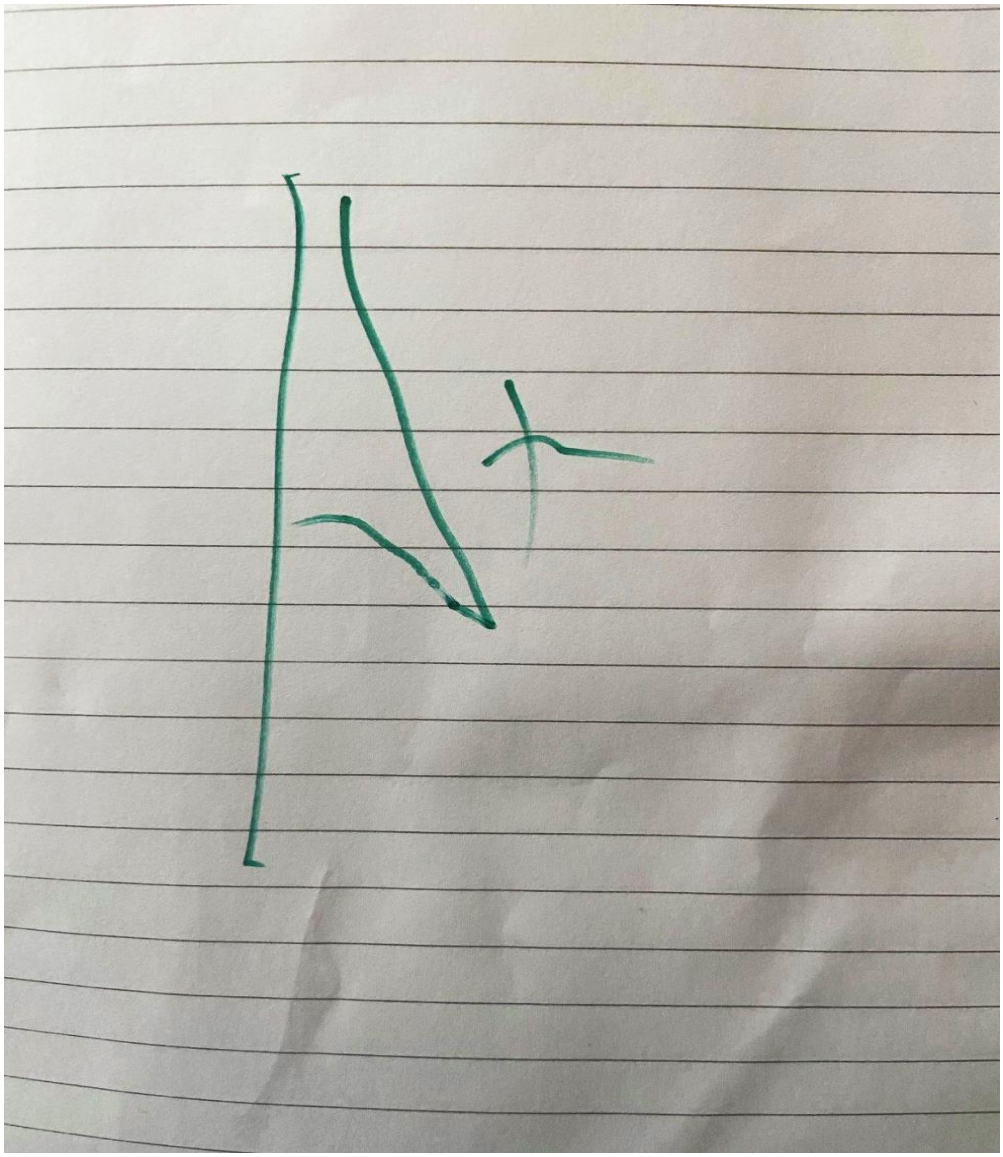
Finally, the study has taught me to find wisdom in my own Resignation, that I have to accept that the story of a thesis has a point of departure, so others can use it 'as a narrative resource' (Frank 2009 p. 190). Like stories, a thesis too must come to an end. Now, it is my responsibility to let these HD stories breathe (Frank, 2010), to have a life of their own. Here in my closing thoughts, I return to voices of the HD participants who after they had read what I wrote about them, gave feedback that ultimately gave me closure, and the feeling that I too can finally let go and breathe.

In their own words, they said…

Daniel: 'You have captured what it's like to live with HD, you caught a lot of essence, growing up with HD and with mum. Thank you.'

Martin: 'I have no problems with it, you listened to what we said, and to what we have to put up with. You wrote it down quite as honest as you could, which is good'.

And in the case of Jen, whose verbal communication had deteriorated, she wrote to say…



A+ (A plus).

Figure 26. **Jen's response**

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APPENDICES

Appendix A Scoping review supplementary

Table 1.A. SPIDER (Cook et al. 2012) table of scoping review study inclusion and exclusion criteria

		Inclusion criteria	Exclusion criteria
S	*Sample*	• Adults diagnosed with HD	•Adults with other diagnosis
PI	*Phenomenon of Interest*	• Studies exploring the lived experiences of HD	• Topics not relevant to lived experiences of HD • Studies that focused on health professionals, families, and caregivers without the perspectives of patients. • Studies that focused on specific topics, interventions or evaluating treatments (i.e. drug efficacy, rehabilitation, RCTs).
D	*Design*	• Qualitative or mixed-methods studies reporting primary qualitative data	• Studies reporting only quantitative data
E	*Evaluation*	• Qualitative analysis of HD experiences	• Evaluation using quantitative methods only
R	*Research Type*	• Peer-reviewed journal articles • Articles written in English	• Systematic reviews, protocols, conference papers, book chapters, thesis and dissertations

Table 2.A Example of scoping review search query

The following search query was used in Scopus* (Updated in August 2019):

(TITLE-ABS-KEY ("Huntington's Disease" OR "HD")) AND (TITLE-ABS-KEY ("Patient experience" OR "liv* experience" OR meaning OR perception OR "patient perspectiv*")) AND (TITLE-ABS-KEY ("Qualitative method*" OR "Mixed method*" OR phenomenolog* OR "grounded theory" OR ethnography OR narrative* OR descriptive OR discourse OR "content analysis" OR "thematic analysis" OR "personal construct theory")) AND (TITLE-ABS-KEY (interview* OR "focus group" OR observation* OR "case stud*"))

*Search terms were adopted in other databases.

Table 3.A Characteristics of included studies in the scoping review

Study, year, country	Qualitative Method	Participants	Sampling and Settings	Aim of study
Zarroti, et al. (2019) UK	Semi-structured interviews Thematic analysis	8 people with early HD (M=3,F=5) Mean age: 50.9 ± 4.7 Mean duration of illness: 6.1 ± 1.45 HD stages: Early=4 Moderate= 4	Sampling: Purposive (postal invite to members of HDA UK) Setting: not stated	To explore the perspectives of people with HD communication abilities
Haplin, M. (2018) USA	Semi- structured face-to-face, and telephone Interviews Grounded theory and thematic analysis	24 people: HD diagnosis (n=10), caregivers (n=14) M=13 F=11 HD stages: not stated	Sampling: Purposive (recruitment aided by social worker and network sampling in HD support groups and retreats) Setting: not stated	To explore the lived experience of HD shaped by genetic discourses and technologies.
Hagen, N. (2018) Sweden	Semi-structured interviews, Interpretative phenomenological analysis (IPA)	11 people: HD diagnosis (n=2), gene positive (n=1), gene negative (n=2), at risk (n=1), families not at risk (n=5) HD stages: not stated	Sampling: Purposive (advertisements in websites aimed to HD community and personal recruitment in HD meetings) Setting: participant's choice of place	To explore the intersections between genes, the body and the lived experiences of a genetic disease.

Maxted, et al (2014) UK	Semi-structured interviews, Interpretative phenomenological analysis (IPA)	7 family dyads (person with HD or at risk and their mother/father/daughter/son) HD stages: not stated	Sampling: Purposive (advertisements in local paper, HD websites support group and discussion forum) Setting: private meeting rooms in public spaces near participants home	To explore the experiences of dyads within HD affected families
Ho and Hocaoglu (2011) UK	Semi-structured interviews, methodology not stated	31 people with HD, 67.7% females, 32.3 % males, 61.3% aged between 30-59 yrs, 38.7% between 60 to 89 yrs. Shoulson and Fann TFC stage: Pre-HD (n=3) Stage 1 (n=1) Stage 2 (n=5) Stage 3 (n=3) Stage 4 (n=9) Stage 5 (6)	Sampling and mode of recruitment: not stated Setting: participant's home	To examine how often people raised issues and concerns regarding impact of HD on everyday life.
Hartelius et al (2010) Sweden	Focus groups and individual interviews, Thematic analysis	28 people consist of HD diagnosis (n=11, M=4, F= 7,) mean age: 50.36 ± 7.67), Family members (n=7, M=3, F=4) Professional carers (n=10, M= 3, F=7) HD stages: Phase 1-2: (n= 6) Phase 3: (n=5)	Sampling: Purposive (invitation to local Huntington members at the hospital) Setting: activity centres, workplace or participant's home	To explore how communication is affected in HD represented by persons with HD, families, carers, and hcps
Schwartz, R (2010) USA	Interviews, Narrative content analysis	10 people with HD, M=3, F=7 Mean age: 48 (range 20-74) HD stages: early (1 year after diagnosis)	Sampling: Purposive (mode of recruitment not stated) Setting: participant homes, telephone	To explore the meaning of being diagnosed with HD

Etchegary, H (2011) Canada	Interviews, IPA	24 people with different test results Positive (n=3), Negative (n=5) Intermediate* (n=2), Tested, unknown results (n=2) HD affected (n=2) Family at risk (n=6) Family, not at risk n=4) HD stages: not stated *results may or may not manifest HD	Sampling: Purposive (recruitment in HD clinics, support groups and newsletter) Setting: participants' home, researchers' office, telephone	To explore living at risk of HD

Table 4.A Study findings and themes from sampled studies

Authors	Findings and/or themes
Zarroti, et al. (2019)	How HD directs and mediates communication Regaining control to improve communication Emotional outflows into communication and the struggle for separation Sheltering as a way to boost confidence in communication.
Haplin, M. (2018)	Conceptualizations of heredity: Guilt, responsibility, and genetic inheritance Pre-symptomatic testing: Chance, uncertainty and genetic testing Disease trajectory: Ambiguity and genetic onset Prognosis: Fatalism and genetic prognosis.
Hagen, N. (2018)	Noticing symptoms in everyday life Neither health nor disease.
Maxted et al (2014)	A spectre hanging over us: HD as a presence within the family Us against the world: Protection, knowledge and control That could be me in 50years: Cyclical changes in identity and role
Ho and Hocaoglu (2011)	The physical/functional theme was strongly present at stage 1, rising steadily and peaked at stage 5. The emotional, social and self-themes were present at all stages and maintained. Financial and legal themes were rarely mentioned and remained

	similar across all stages. The cognitive theme was featured in stage 1 and 4, but hardly present at pre-clinical stage and stage 5.
Hartelius et al (2010)	Communication has changed Factors that influence communication negatively Factors that influence communication positively.
Schwartz, R (2010)	Discovering the existence of HD Confirming the diagnosis of HD Revealing the diagnosis to others Experiencing the reverberations of HD
Etchegary, H (2011)	Biological disruption, including threats to self-identity and changes to relationships with others Zones of relevance, the conditions under which risk is or is not salient

Table 5.A Scoping review protocol on qualitative synthesis of the illness experience of HD

Review question To systematically identify qualitative research that investigates the illness experience of people with HD To generate a thematic synthesis of the themes evident in existing qualitative literature
Searches Searches will be undertaken in electronic databases: MEDLINE, CINAHL, PsycINFO, SCOPUS and WEB of SCIENCE. The search strategy will run from the earliest date available until the search date. The search terms with MeSH heading and free text will be the following: 1. 'Huntington's Disease' OR 'HD' 2. 'Patient experience' OR 'liv* experience' OR meaning OR perception OR 'patient perspectiv*' 3. 'Qualitative method*' OR 'Mixed method*' OR phenomenolog* OR 'grounded theory' OR ethnography OR narrative* OR descriptive Or discourse OR 'content analysis' OR 'thematic analysis' OR 'personal construct theory' 4. Interview* OR 'focus group' OR observation* OR 'case stud*' To maximise the search results, reference list screening of relevant studies, snowballing and hand searching will be utilised. Support from academic liaison librarian will be obtained, as well as contact with key researchers in the field.
Types of study to be included

Inclusion: Published qualitative studies. Mixed methods will be included if the qualitative methods meet the inclusion criteria and informs a substantial part of the study. To maximise identification of reports not included in databases, grey literature will be included. This will be identified through searches of relevant charities websites (Neurological Alliance, Huntington's Disease Association) and grey literature resources (OpenGrey, PsycExtra, and the British Library's EThOS database).

Exclusion:

- Quantitative studies
- Studies written in non-English language

Condition or domain being studied

Huntington's disease

Participants/population

Inclusion: People with HD, and their families or caregivers.

Exclusion: People with other conditions than HD.

Interventions/exposure

Inclusion: Qualitative studies exploring views and experiences of people with HD.

Exclusion:

- Studies that involve people with diseases other than HD
- Studies that involve people at risk of HD without clinical diagnosis
- Studies that focus on health professionals, families and caregivers without the perspective of people with HD.

Main outcome(s)

To provide an insight on the views and experiences of people with HD.

This will provide implications for clinical practice and inform research priorities.

Additional outcome(s)

To identify gaps in HD qualitative literature.

Data extraction (selection and coding)

Using the search strategies previously outlined, articles will be selected based on PRISMA guidance*, duplicates and irrelevant studies will be removed. Titles and abstract will be screened for eligibility based on inclusion criteria. Any eligibility for inclusion will be addressed by full text screening, and missing data will requested from study authors.

*Reference: Moher D, Liberati A, Tetzlaff J, Altman DG, The PRISMA Group (2009). *Preferred Reporting Items for Systematic Reviews and MetaAnalyses: The PRISMA Statement. PLoS Med* 6(7): e1000097.

Risk of bias (quality) assessment

The Critical Appraisal Skills Programme (CASP)* tool for quality assessment will be applied in this study.

*Reference: CASP-uk.net. (2019). [online] Available at: *https://casp-uk.net/wp-content/uploads/2018/01/CASP-Qualitative-Checklist-2018.pdf* [Accessed 6 Feb. 2019].

Strategy for data synthesis

The analysis and synthesis will be divided into three stage thematic synthesis as proposed by Thomas and Harden 2008*. All of the 'results' or 'findings' of the studies will be used for qualitative analysis, with text entered verbatim in NVivo11 software for coding.

The first stage will be the inductive line by line coding and organisation of new codes of related areas of each study. The second stage will develop descriptive themes by the process of identifying resemblances and disparities between the initial codes, and further grouping into categories. The final stage will be generating analytical themes that go beyond the initial synthesis of the original study findings, the use of descriptive themes will aim to answer the focus of the systematic review question.

Analysis of subgroups or subsets

None planned

*Reference: Thomas, J. and Harden, A. (2008) Methods for the thematic synthesis of qualitative research in systematic reviews. *BMC medical research methodology,* 8 (1), 45.

Anticipated or actual start date

04.11.2019

Anticipated completion date

05.10.2020

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Appendix B Quality assessment of PLTNC narrative studies and studies that referenced Arthur Frank's typology

Critical Appraisal Skills Programme (CASP)

Accessed: 10th December 2018 https://casp-uk.net/

Screening Questions

1. Was there a clear statement of the aims of the research?

   What was the goal of the research? Why was it thought important? Its relevance.

2. Is a qualitative methodology appropriate?

   Did the research seek to interpret or illuminate the actions and/or subjective experiences of research participants? Is qualitative research the right methodology for addressing the research goal.

Detailed Questions

3. Was the research design appropriate to address the aims of the research? Has the research justified the research design?

4. Was the recruitment strategy appropriate to the aims of the research?

   Did the researcher explained how the participants were selected? Did they explain why the participants they selected were the most appropriate to provide access to the type of knowledge sought by the study? Was there any discussions around recruitment?

5. Was the data collected in a way that addressed the research issue?

   Was the setting for data collection justified? Was it clear how data was collected? Did the researcher justified the methods chosen? Did the researcher has made the methods explicit. Was the form of data clear?

6. Has the relationship between researcher and participants been adequately considered?

   Did the researcher critically examine their own role, potential bias, and influence. How the researcher responded to events during the study and whether they considered the implications of any changes in the research design.

7. Have ethical issues been taken into consideration?

Is there sufficient detail of how the research was explained to the participants. Did the researcher discuss the issues raised by the study (informed consent/ confidentiality)?

8. Was the data analysis sufficiently rigorous?

   Is there an in- depth description of the analysis process? Is thematic analysis used- if so, is it clear how the themes were derived. Is there sufficient data presented to support the findings? Did the researcher critically examine their own role, potential bias, and influence.

9. Is there a clear statement of findings?

   Are the findings explicit. Is there adequate discussion of the evidence both for and against the researchers' arguments. Does the researcher discuss the credibility of the findings? Are the findings discussed in relation to the original research question?

10. How valuable is the research?

    Does the researcher discuss the contribution the study makes to existing knowledge or understanding? Does the researcher identify new areas where research is necessary? Does the researcher discuss whether/how the findings can be transferred to other populations?

Table 1.B Selected narrative studies of progressive long-term neurological conditions (PLTNC)

Study title, country, author, year	Phenomenon of interest	Study aim, design and methods	Study findings	Comments, limitations, and study relevance to HD	CASP Score
Multiple Sclerosis in its context: Individual narratives (USA) Thapar et al. (2001)	MS	To illustrate how the meaning of geographic activity space is dependent upon context (MS and their broader social setting). In this study, activity space refers to frequent sites visited for social, occupational and leisure activities. Two cases were selected from a larger study of MS in-depth interviews (n-53) that focused on activity spaces; disease impact and issues on spatial negotiation. Cases are both males, 38 and 55 years old. One rated with moderate, and the other high level of disability based on Environmental Status Scale. The scale provides assessment on seven items of work, finances, personal assistance, transport, community services and social activities.	The study highlights that for both persons with MS, the activity space was not defined by spatial parameters but through the richness of social-spatial content and its meaning in terms of human interaction. The same disease (MS) has a widely different meaning in lives of individuals living in the same physical activity space (North-eastern Ohio)	The two cases result in different social-spatial outcomes due to the different combinations of their contextual factors the personal outcomes are determined by different social-spatial factors such as individual, social support, economic, cultural and environmental.	7
Story making and Storytelling: making sense of living with Multiple Sclerosis (New Zealand)	MS	To understand the lived experience of women with MS using symbolic interactionism. Narrative quotes were gathered from women (n-16) using focus group (n-6) and interviews (n-10).	Analysis of narrative data generated five symbolic and theoretical codes that suggest women interpret MS being an 'aggressor', a 'saviour', a 'partner', a 'guest' and an 'adversary. 'The study highlighted storytelling and story making demonstrated a means of constructing	Making sense of things (through narratives) enabled a sense of transformation of how life is lived in presence of chronic illness (such as MS). The study also discussed the use of metaphors as to express meaning when ordinary words seemed inadequate.	9

Wright-St.Clair (2003)			and recon structuring meaning and life events.	The interview format stated a semi-structured approach, with topics around self and relationship with others. It would be useful to get insight on the examples of these. The study did not elaborate on the narrative analysis and steps taken address validity and reflexivity.	
Two women with multiple sclerosis and their caregivers: Conflicting normative expectations (Netherlands) Abma et al (2005)	MS	To describe and illustrate Walker's expressive-collaborative view of morality to interpret the normative expectations of two women with multiple sclerosis. Two case studies from a larger project of MS patients (n-15) and their caregivers (n-24) A 39 and 48-year-old women, with average MS diagnosis of 20 years.	Walker's method also helps professionals to understand the uniqueness of patients. The two narratives show that, although the women had a similar MS diagnosis, the meaning with which their situation was endowed was very different, and also need different care. The study argued that there is a tendency in health care to prescribe the use of protocols and health care standards. The results show that these care recommendations must be tailored according to the individual needs of patients. Walker's framework challenges health care professionals to place standard ways of thinking in a contextual perspective.	The specific needs of patients can be revealed through interaction and communication. Listening and understanding is very important but often hard to practice for health professionals. Analysis used Lieblich et al (1993) approach with emphasis on 'member checking' and 'inter-rater reliability' to enhance verification of the findings. It is unclear what the author refers to as 'natural' conversation.	10

How people with motor neuron disease talk about living with their illness: a narrative study (United Kingdom) Brown and Addington-Hall (2007)	MND	To explore the experiences of living and coping of people with MND leading to the disease progression. Thirteen adults participated in a longitudinal interview (eighteen months, three months interval) in narrative case studies design.	Four storylines or types of narratives were identified: Sustaining (living well, keeping active and engaged), Enduring (feeling disempowered, unable to fight for life or against death), Preserving (survival) and Fracturing (loss and fear of what is to come)	Storylines can help people with complex narratives make sense of their experience and serve as organising threads to help patients, families and HCPS better understand the illness experience. The study recognised that different disease stages or symptoms were not examined which could have influenced the storylines.	10
Narrative as snapshot: glimpse into the past in Alzheimer's discourse (USA) Hamilton (2008)	AD	To examine the intersection of narrative, identity and memory in AD by in-depth examination of linguistic construction. One case study of a women in her 80's with moderate to severe dementia. The study was conducted over 4.5 years with the total of five interviews (2 hours, 30 minutes).	The narrative choices can reveal much about how narrators see themselves and how they wish to be seen by others. Identity construction can be understood in the limitation of coherent reconstructions of the past.	The analysis draw heaving on linguistic discourses drawing upon the frameworks of Labov (1972) and Chafe (1994). Challenges have been emphasised on the study of linguistic construction with conditions such as AD. The emphasis on the 'person' and using personal objects to facilitate conversation can evoke well-being and positive interaction despite incoherent narratives.	8
Narrative and identity in Alzheimer's disease: A case study (Sweden)	AD	To illustrate how persons with AD use available linguistic and cognitive abilities with non-verbal aspects to communicate and negotiate identity.	The results of narrative analysis suggest that other aspects than temporal and referential organisation had become an important resource for this case. For people diagnosed with cognitive and linguistic impairments such as AD, their	People with AD try to use and invent alternative communicative resources to maintain sense of self and identity.	8

Hyden and Orulv (2008)		One case study of a woman, who was diagnosed for 5 years, her age was not stated. She currently resides in the elderly care home. The study was conducted over a period of 5 months in the care setting, involving other residents, visitors and care staff. The focus was one story of one woman told several times in different occasions.	discontinuous narratives does not affect continuous self and identity, which can be a problem to people without these impairments.	The study illustrates that for researchers studying people with similar diagnosis to focus their analysis on function and various discourses in the interaction.	
Ripples from a stone skipping across the lake: a narrative approach to the meaning of Huntington's disease (USA) Schwartz (2010)	HD	To explore the meaning of being diagnosed with HD. Ten participants (seven women, three men) within the first year of diagnosis.	An integrated narrative, "The Story of HD: Ripples from a Stone Skipping across the Lake," was created from participant stories. The stories were analysed for plot, predicaments, protagonist, and antagonist. The predicaments of "discovering the existence of HD," "confirming the diagnosis of HD," "revealing the diagnosis to others," and "experiencing the reverberations of HD" served as the main chapters that formed the structure of the stories. Narrative analysis using holistic-content perspective, the outcome of the analysis was presented in an integrative narrative.	The psychological of HD diagnosis has implications for patients and their extended families. The study suggests that nurses should develop their understanding of the role of genetics and develop a more comprehensive role to assist patients and families in finding the personal meaning of being diagnosed with HD. The author outlined several challenges in narrative interviewing, such as some participants refusal to be audio-recorded and difficulties with transcription, however, did not state the steps to overcome or resolve these barriers or how these affected the analysis and study findings. Regardless, the important insights provided clinical implications and importance of use of narratives to better understand diagnosis of the illness.	9
Stories about life narrated by people	AD	To explore how people with AD present their life story.	Themes of contentment, connectedness, self-reliance and personal growth were identified as core dimensions in the	The study reinforces that use of narrative approaches such as life story and storytelling can	10

with Alzheimer's disease (Sweden) Karlsson et al (2014)		Nine participants (five women and four men), aged 60-81 years old in different disease stages participated in the study conducted within 6 months.	participants' life stories. The study acknowledges that people with AD can have a positive hopeful approach in life, and the use of life story work can help support their sense of self and identity.	be support and sustain identity situated in the social, cultural and historical context. However, the study utilised pre-defined areas in their interview questions that might have influenced the generated themes.	
'There was no great ceremony': patient narratives and the diagnostic encounter in the context of Parkinson's (United Kingdom) Peek (2017)	PD	The study draws on stories of diagnosis from a broader narrative study on the lived experience of PD. Thirty-seven adults with PD (sixteen women and twenty-one men) aged from 29 to 78 and received diagnosis from 3 months to 33 years.	Three key concepts connected the participant's diagnostic encounter: a 'bareness' or lack of 'ceremony', a sense of emotional and physical 'abandonment' and the impact on a person's illness story when faced with a 'hierarchy' of illness. The participants' narratives which included a structured account of experience rather than factual record signifies the individual meaning of their experiences.	The study suggests many participants felt that emotional impact of PD diagnosis remained unacknowledged. Regardless who gives the diagnosis, clinicians need to adapt consultation style with open dialogues and acknowledge the 'human significance' of the diagnostic encounter. The study acknowledged the importance of patient narratives in understanding the illness experience, more so in the context of receiving a diagnosis. A safe environment is needed to share best practice which involves the participation and views of patients and clinicians.	10
Understanding Parkinson's through visual narratives: "I'm not Mrs. Parkinson's" (Canada)	PD	To deepen understanding of the experience of living with PD and its implications for occupation through a narrative visual methodology (photo-elicitation).	Three major themes were identified: Framing the meaning of PD (accepting the disease as part of who they were); Negotiating engagement in occupation (ongoing deliberation whether to continue engaging in certain aspects of	Participants' narratives highlighted the centrality of occupation in day-to-day life. The study used visual narratives to examine interrelationship of identity and occupation in PD,	10

Lutz et al (2018)		Six adults with PD, (two females, four males, aged between 57-73 years old, within 5 to 10 years of diagnosis) Participants were asked to take photographs and share verbal narratives to illustrate their experience of living with PD.	life as PD progressed); and Being ready to accept changes that impact personal or social identity (Readiness to accept help and to identify as someone with PD).	stories of engagement in occupation were linked to sense of identity. The participants were at the similar progression of disease stage and older adults (mean age 65), a diverse range of participants could yield a different understanding.	

Table 2.B CASP Assessment of selected PLTNC narrative studies

Author (Year)	Was there a clear statement of the aims of the study?	Is qualitative methodology appropriate?	Was the research design appropriate to address the aim of the research?	Was the recruitment strategy appropriate to the aims of the research?	Was the data collected in a way that addressed the research issue?	Has the relationship between researcher and participants been adequately considered?	Have ethical issues been taken into consideration?	Was the data analysis sufficiently rigorous?	Is there a clear statement of the findings?	Total no. of yes in Yes in the final quality score
Thapar et al (2001)	YES	YES	YES	YES	NOT SURE	NO	The authors did not provide any ethical statement	YES	YES	7
Wright-St Clair (2003)	YES	YES	YES	YES	YES	The author did not state relationship or position in the study	YES	YES	YES	9

Abma et al (2005)	YES	YES	YES	YES	YES	YES	YES	YES	YES	10
Brown and Addington-Hall (2007)	YES	YES	YES	YES	YES	YES	YES	YES	YES	10
Hamilton (2008)	YES	YES	YES	YES	YES	Not stated	The author did not provide ethical considerations	YES	YES	8
Hyden and Orulv (2008)	YES	YES	YES	YES	YES	Not stated	No ethical statement provided	YES	YES	8
Schwartz (2010)	YES	YES	YES	YES	YES	YES	Can't tell	YES	YES	9
Karlsson et al (2014)	YES	YES	YES	YES	YES	YES	YES	YES	YES	10
Peek (2017)	YES	YES	YES	YES	YES	YES	YES	YES	YES	10
Lutz et al (2018)	YES	YES	YES	YES	YES	YES	YES	YES	YES	10

Table 3.B Example of PLTNC search strategy

The following search query was used in Scopus (October 2019):
(TITLE-ABS-KEY (“Parkinson's Disease” OR “PD")) AND (TITLE-ABS-KEY ("Patient experience" OR "liv* experience" OR meaning OR perception OR "patient perspectiv*")) AND (TITLE-ABS-KEY ("narrative method*" OR "narrative inquiry" OR “narrative*” Search terms were adopted in other diseases which includes Multiple Sclerosis (MS), Motor Neuron Disease (MND) and Alzheimer’s Disease (AD).

Table 4.B Selected studies that used Arthur Franks (1995) illness typology

Study title, country, author, year	Phenomenon of interest	Study aim, design, and methods	Study findings	Comments, limitations, and study relevance	CASP Score
Illness narratives: time, hope and HIV. (Australia) Ezzy (2000)	HIV	To determine how people living with HIV/AIDS make sense of their illness experience. Mixed methods study: quantitative survey (n=914) and qualitative interviews (n=45). For the qualitative aspect, the interview questions were centred around employment, religion and understanding of the future. Data were	Three dominant narrative types were established: 1. Linear restitution 2. Linear chaos 3. Polyphonic Linear restitution narrates life as normal continuation of goals and values, despite diagnosis. Uncertainty	The study results were driven by the quantitative results and the established narratives were derived basically from participants quotes. Despite the lack of context of the narratives, a useful discussion was provided on the narrative types. Overall, there was strong preference for linear narratives. However, the author argued that Polyphonic narratives provide the person a	8

		analysed using grounded theory and narrative theory.	about the future were resolved and hope is placed on medical interventions and advances in treatment. Linear chaos is living in an empty present and bleak future with experiences of depression, anger and isolation. Polyphonic are contradictory stories and voices, and belief that future can be unpredictable.	greater flexibility to adapt to the uncertain future.	
Understanding breast cancer stories via Frank's narrative types. (Canada) Thomas-MacLean (2004)	Breast CA	To utilise Frank's narrative types in a phenomenological study to enhance understanding of embodiment after breast cancer. Data collected though one focus group (n=5) and two in-depth interviews (n=12). Analysis of transcripts included both thematic and narrative approaches.	Restitution is the most desired narrative, while Chaos is prominent in certain situation such as when they speak of bodily movements and other people's involvement. Quest is least represented and occupies a more tenuous place in the analysis.	The author recognised the difficulty of participants articulating their experiences due to the long-term effects of cancer and the characteristics of treatments. None of the participants undergone reconstructive surgery and could affect the strong desire for restitution. The use of Frank's typology was an accessible device to understand complex experiences and uncover narrative forms.	10
Quest, chaos and restitution: Living with chronic fatigue syndrome/ myalgic encephalomyelitis (UK).	Chronic fatigue syndrome (CFS)/ myalgic encephalomyelitis (ME).	To explore how people with CFS/ME construct their illness experience. Seventeen interviews (11 women and 6 men) for up to three sessions were conducted. Participants were asked to narrate their illness experience from onset of symptoms to present	The study proposed a trajectory of narrative typology starting with restitution, moving to chaos, then most were back to restitution and on to quest. The study contrasted the unstable restitution findings of Ezzy (2000) with HIV/AIDS and Thomas-Maclean (2004) on Breast CA, that people with CFS/ME do not actively	The study established a narrative pattern and concluded that CFS/ME illness experience fits into a possible trajectory. While Frank's typology fits in the narratives, the lack of contextual details of the participants makes the Quest illness, 'too clean'. No study implications were discussed.	10

Whitehead (2006)		day. Frank's illness typology was used in the data analysis.	embrace restitution for they are never symptom free.		
How people with motor neurone disease talk about living with their illness: a narrative study (UK) Brown and Addington-Hall (2008)	MND	To explore how MND patients construct their illness experience Narrative case studies of thirteen adults with MND. Longitudinal interviews were conducted at three months interval over 18months. Analysis focused on form and content with the use of Frank's established illness storylines.	The study identified four storylines: 1. Sustaining 2. Enduring 3. Preserving 4. Fracturing Sustaining is about actively living well as possible, while in contrast to Enduring, unable to fight for life and feeling disempowered. Preserving strives for survival while Fracturing concerns with loss and fear of the future.	The findings suggest that storylines do not suggest people's characters or stages of illness rather can be used to organising narrative threads from disordered experiences. The study provided general implications in understanding MND and placed emphasis on the therapeutic values of listening to illness experience.	10
Now let me tell you in my own words: narratives of acute and chronic low back pain (USA) Vroman et al (2009)	Low Back Pain	To examine the broader experiences of living with acute and chronic low back pain (LBP) A part of wider study of 143 participants the qualitative aspect was derived from the questionnaire with an open-ended question asking participants to talk about their LBP, in their own words.	The study found two themes: 1. Challenges to LBP authenticity 2. Consequences of LBP, which has led to life disruptions to physical limitations or emotional distress. The narratives were solely told in chaos.	The authors found that the structure of the written narratives was described in chaos, devoid of hope, full of frustration, anger, and blame. This was perhaps influenced on how the written question was framed, and the question was at the end of a lengthy questionnaire. Which stated: 'If you could talk to researchers, what would you like them to know about LBP?'	10

		Thematic content and structural analysis were conducted using Frank's typology.		A detailed exploration of the narratives could possibly yield other types, than just chaos.	
Do Men's and Women's Accounts of Surviving a Stroke Conform to Frank's Narrative Genres? (UK) France et al (2013)	Stroke	To explore men and women's life after stroke using Frank's illness storylines. Taken from a larger sample, secondary analysis of eighteen (9 women and 9 men) interview transcripts were conducted. Transcripts were transferred to NVivo and consequently were to 'notes' corresponding to Franks' narrative genre.	The study found that gender has no influence on the narrative type, but rather largely influenced by severity, degree of anticipation or actual recovery. Most accounts have overarching narrative genre, the most common was quest memoir.	An interesting study that used stories 'as coded materials to be analysed'. The authors claimed that the analysis operated a 'replicable approach to analyses.' These claims were in contrast to the reproducibility of narrative studies (Reissman 2008) and the Frank's theoretical framework of dialogical approach to analysis and interpretation (Frank 2010).	8
Quest, chaos and restitution: The illness narratives of individuals diagnosed with fibromyalgia syndrome (UK). Diver et al (2013)	Fibromyalgia	To describe the experiences of people recently diagnosed with Fibromyalgia using Frank's illness typology. Twenty-three individuals (22 females, one male) were interviewed on three occasions and data were analysed using narrative thematic content.	The study found that illness experiences of people with Fibromyalgia conformed to three dominant typologies of Quest, Chaos and Restitution. The findings propose two subdivisions of Quest: as active engagement and active disengagement.	The study claims the robustness of Frank's theoretical model and has expanded the narrative typology of quest. Although the narratives were derived from 23 participants, the presentation of four vignettes provided the breadth and key features of the narrative findings. This illustrated Frank's claim that narrative is best represented by a study of the particular.	8

A qualitative study examining the illness narrative master plots of people with head and neck cancer (UK) Reid and Soundy (2019)	Head and Neck CA (HNC)	To identify the most supported narrative plots from diagnosis to a year post treatment of people with HNC. Eighteen people (12 males and 6 females) were purposely recruited a hospital-based cancer centre and participated in a single interview. The methodology of hermeneutic phenomenology, the methods of Q sort and structural narrative analysis of master plots were used.	The study identified five illness narrative master plots: 1. The responsive and reflective narrative 2. The frail narrative 3. The recovery narrative 4. The survive or die narrative 5. The personal project narrative	The study identified alternative narrative plots to Frank's (1995) typology and that could enhance understanding on how people with HNC perceive their experience. The authors acknowledged that confounding variables or key demographics may influenced the narration of master plots. The study provided generic and specific guidelines that could be useful for health professionals and applicable to other chronic illness.	10
Finding oneself after critical illness": voices from the remission society (Norway). Ellingsen et al (2021)	Patients who been admitted to critical care units	To explore how former critically ill patients craft and recraft their illness narratives. Nine participants (four women and five men) participated in two focus groups and one individual interview. A phenomenological approach to analysis was used leading to narrative structure based on Frank's Quest illness typology.	Th study findings implicate that participant's quest to find oneself after critical illness, forcing new understanding of themselves. The study further suggests the need for recognition and support through a critical illness trajectory and listening to patient's stories can be vital for recovery.	The study was heavily drawn by theoretical concepts informed by Heidegger (1962) and Frank (1995). It wasn't clear how individual narratives were obtained and or the lack of information on the number of people participated in focus groups and individual interviews respectively. The study place emphasis on listening to the patient story and the use of phenomenology as a useful approach to provide tailored patient care.	9
Long Covid – The illness narratives (UK)	Covid-19	To explore the rapid emergence of long Covid and the unique status of their illness narratives.	The study findings described the participants narratives and key features of their stories which created a persuasive account of a new symptom best with setbacks and	The study argued that individual stories make little sense however collectively it presented a rich description of a new illness that needs better recognition by care professionals and health systems.	9

Rushforth et al (2021)		114 UK based participants participated in narrative interviews and focus groups. Data were inputted to NVivo12 for coding and thematic analysis. The theoretical framework were informed by Frank's socio-narratology (2010) , Mattingly's therapeutic emplotment (2010) and Bakhtin's polyphonia (1984)	unrecognised by health professionals. The study found that most unique feature of long Covid was the absence of health professionals to witness them and rather largely visible accounts were found in online communities and social media.	A very large sample study for qualitative research. The authors stated they used thematic analysis, however it appeared to be more of content analysis. The study has a more quantitative feel to it, with very little quotes or contextual information to support the narratives.	

Table 5.B CASP Assessment of selected studies that referenced Arthur Frank's illness typology

Author (Year)	Was there a clear statement of the aims of the study?	Is qualitative methodology appropriate?	Was the research design appropriate to address the aim of the research?	Was the recruitment strategy appropriate to the aims of the research?	Was the data collected in a way that addressed the research issue?	Has the relationship between researcher and participants been adequately considered?	Have ethical issues been taken into consideration?	Was the data analysis sufficiently rigorous?	Is there a clear statement of the findings?	Total no. of yes in Yes in the final quality score
Ezzy (2000)	YES	YES	YES	YES	YES	NO	The author did not provide any ethical statement	YES	YES	8
Thomas-MacLean (2004)	YES	YES	YES	YES	YES	YES	YES	YES	YES	10
Whitehead (2006)	YES	YES	YES	YES	YES	YES	YES	YES	YES	10
Brown and Addington-Hall (2008)	YES	YES	YES	YES	YES	YES	YES	YES	YES	10
Vroman et al (2009)	YES	YES	YES	YES	YES	YES	YES	YES	YES	10
France et al (2013)	YES	YES	YES	YES	YES	Not stated	No ethical statement provided	YES	YES	8

Diver et al (2013)	YES	YES	YES	YES	YES	Not stated	No ethical statement provided	YES	YES	8
Reid and Soundy (2019)	YES	YES	YES	YES	YES	YES	YES	YES	YES	10
Ellingsen et al (2021)	YES	YES	YES	YES	YES	YES	The authors did not provide ethical considerations	YES	YES	9
Rushforth et al (2021)	YES	YES	YES	YES	NOT SURE	YES	YES	YES	YES	9

Appendix C participant details

Table 1.C Participatory Research Exercise (PRE) participants details

Name	Age	Gender	Other relevant HD information
Persons with HD			
Graeme	53	M	Late-Stage HD, TFC Score 3, diagnosed in 2013
Julie	52	F	Late-Stage HD, TFC Score 2, diagnosed in 2007
Family Caregivers			
Lenny	54	F	Mother of HD patient (juvenile onset)
Tyler	38	M	Son of HD patient, At risk (status undisclosed)
Professional Caregivers/ Health Care Assistants (HCAs)			
Linda	55	F	Working with HD for 6 years
Trisha	43	F	Working with HD for 2 years
Zahid	28	M	Recently working with HD, less than 1 year
Registered Nurses			
Barbara	63	F	Working with HD for 10 years
Jasmine	57	F	Working with HD for 11 years
Cathy	33	F	Working with HD for 6 years

Appendix D. Life Story template

MY LIFE STORY

Date of Birth: _____

Place of birth: _____

MY FAMILY

Mother's Name and Occupation: _____

Father's Name and Occupation: _____

My Siblings:

_____

_____

_____

My Children:

_____

_____

_____

Grandparents or other family members such as aunts or uncles:

_____

_____

_____

_____

_____

MY CHILDHOOD

Write about your significant childhood memories. Example: home life, school, family holidays

MY WORKING LIFE

Write about significant memories about work. Example: first job, favourite job, places of work

SIGNIFICANT RELATIONSHIPS

Mention significant people in your life and your relationship to them. Example: Friends, partners or family

Do you have any favourite pets?

SIGNIFICANT LIFE EVENTS

This may include significant things or events that had a significant impact on your life.

Please include event dates if you can. **(Please only include things that you are comfortable with sharing)**

MY LIFE NOW

What is important to me now? Things I like doing.

PEOPLE WHO ARE IMPORTANT TO ME NOW

Please write down the important people in your life now and your relationship with them.

MY WISHES FOR THE FUTURE

Please write down any future wishes or things that you would like to do. This includes what is important to you.

Appendix E. Example entries on field notes and researcher journal

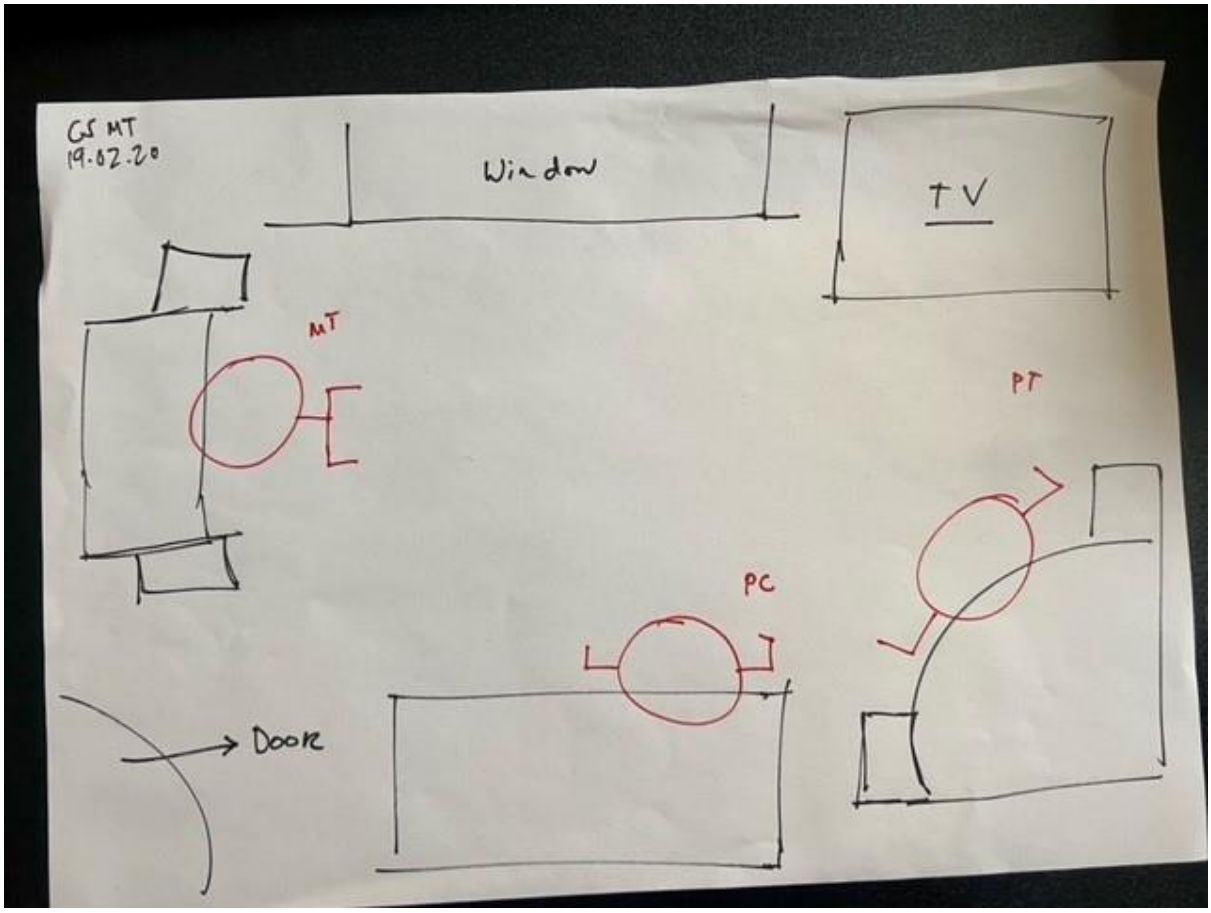
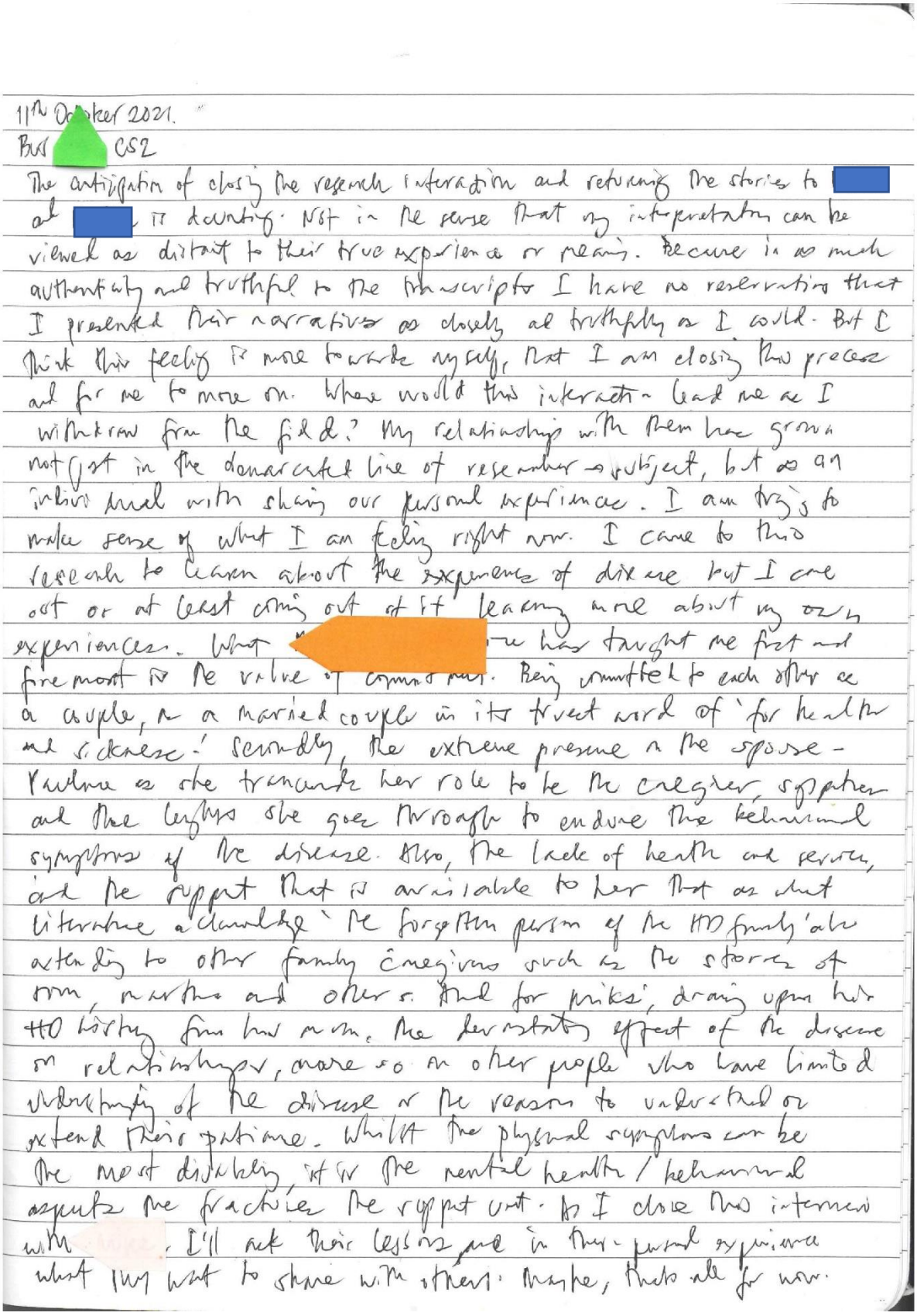


Figure 1.E. Sketch illustration of the room layout and interview setting


11th October 2021.

But CS2

The anticipation of closing the research interaction and returning the stories to [redacted] at [redacted] IT daunting. Not in the sense that my interpretation can be viewed as distant to their true experience or meaning. Because in as much authenticity and truthful to the transcripts I have no reservations that I presented their narratives as closely as truthfully as I could. But I think this feeling is more towards myself, that I am closing this process and for me to move on. Where would this interaction lead me as I withdraw from the field? My relationship with them has grown not just in the demarcated line of researcher-subject, but as an individual with sharing our personal experiences. I am trying to make sense of what I am feeling right now. I came to this research to learn about the experience of disease but I am out or at least coming out of it learning more about my own experiences. What [redacted] has taught me first and foremost is the value of communication. Being committed to each other as a couple, as a married couple in its truest word of 'for health and sickness.' Secondly, the extreme pressure on the spouse - Pauline as she transcends her role to be the caregiver, supporter and the lengths she goes through to endure the behavioural symptoms of the disease. Also, the lack of health and service, and the support that is available to her that as that literature acknowledge 'the forgotten person of the HD family' also extending to other family caregivers such as the stories of mum, mothers and others. And for Nikki, drawing upon her HD history from her mum. The devastating effect of the disease on relationships, more so than other people who have limited understanding of the disease or the reasons to understand or extend their patience. Whilst the physical symptoms can be the most disabling, it is the mental health / behavioural aspects the fractures the support unit. As I close this interview with [redacted]. I'll ask their lessons and in their personal experience what they want to share with others. Maybe, that's all for now.

Figure 2.E. Example of research journal entry

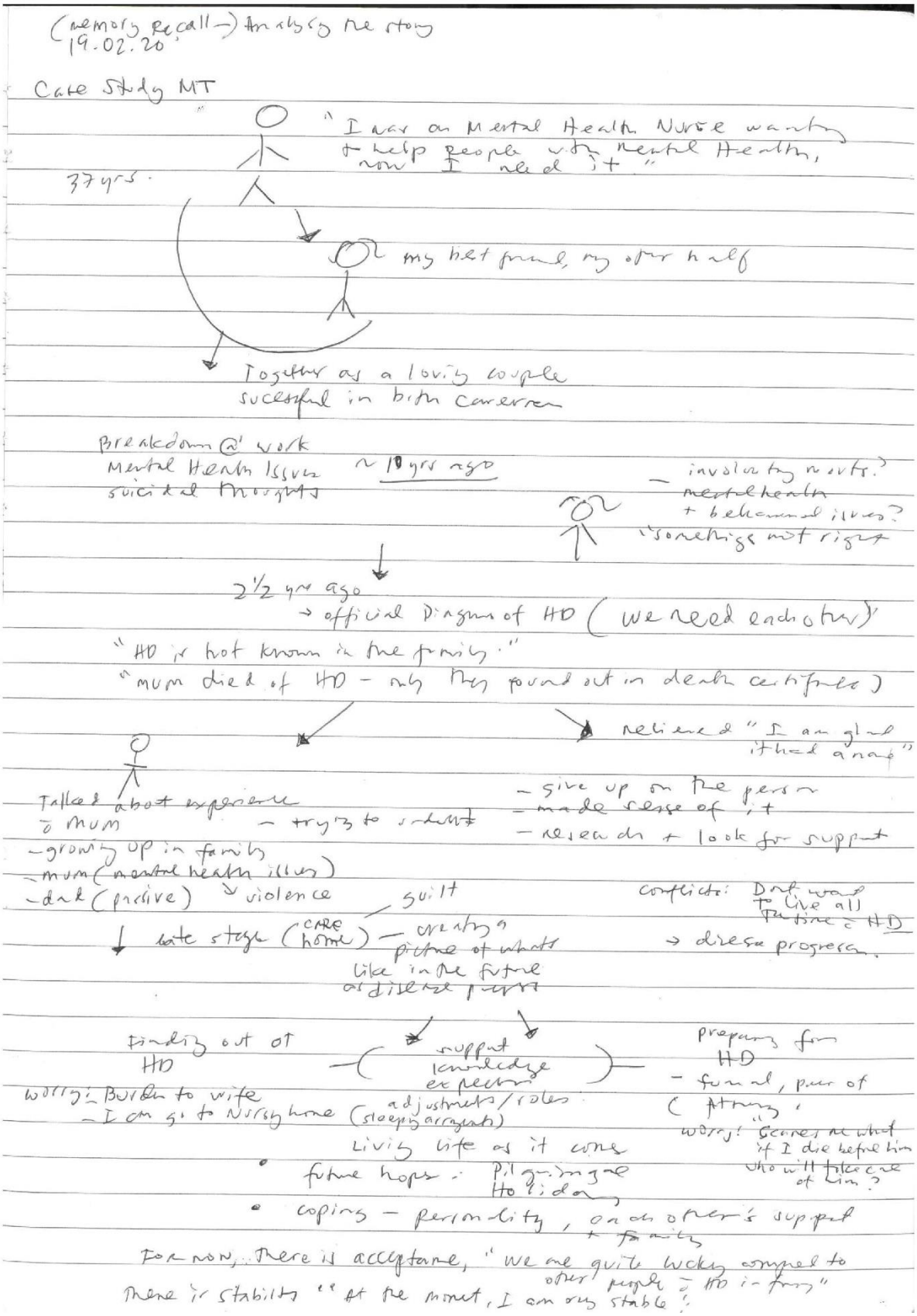
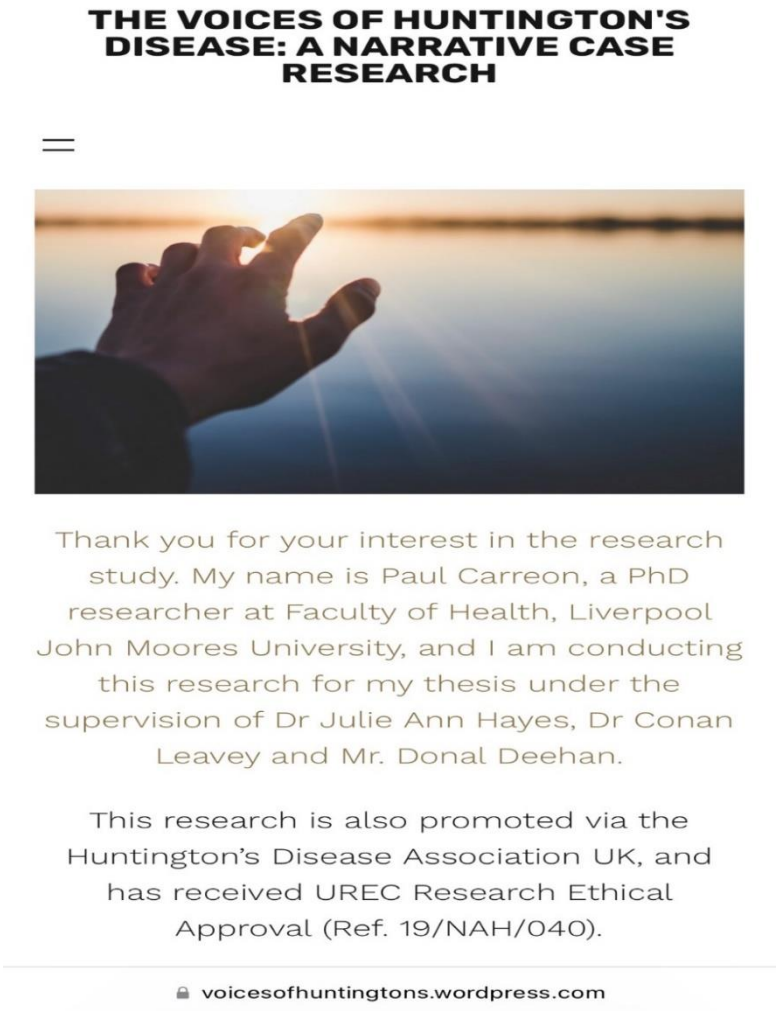


Figure 3.E. Example of field notes entry

Appendix F. Homepage of study website

https://voicesofhuntingtons.wordpress.com/



Appendix G. Study ethical approvals

5th June 2019

With reference to your application for Ethical Approval

Richie Paul Carreon, PGR - The voices of people living with, and affected by Huntington's Disease (HD)

UREC reference: 19/NAH/023

The University Research Ethics Committee (UREC) has considered the above application. I am pleased to inform you that ethical approval has been granted subject to the provisos listed below. Once the final version of the ethics application with the provisos addressed has been emailed to researchethics@ljmu.ac.uk, the study can commence.

(Please note, UREC will not check that the provisos have been applied in the final version of the ethics application and will not email any further approval notifications to the applicant once the final version of the ethics application has been forwarded to UREC. If the applicant does not want to apply the provisos as stated below, the applicant must notify UREC and resubmit the ethics application for further review)

Provisos:

- Given that the participants might have an impairment of, or disturbance to, the functioning of the mind or brain (https://www.hra.nhs.uk/planning-and-improving-research/policies-standards-legislation/mental-capacity-act/) please operate a procedure for the researcher to assess for themselves whether the impairment or disturbance is sufficient that the person is unable to make the decision about participating in the study. For example, in the time periods before the focus group, when informing participants the researcher could presume the individual has capacity and ask them questions to help you decide whether they probably have the capacity to consent to this "minimal risk" study - questions that tell you that the individual understands the decision to made about participating in the study, that they retain the information provided to them and they can weigh up the pros and cons of making the decision.

Approval is given on the understanding that:

- any adverse reactions/events which take place during the course of the project are reported to the Committee immediately by emailing researchethics@ljmu.ac.uk;
- any unforeseen ethical issues arising during the course of the project will be reported to the Committee immediately emailing researchethics@ljmu.ac.uk;
- the LJMU logo is used for all documentation relating to participant recruitment and participation e.g. poster, information sheets, consent forms, questionnaires. The LJMU logo can be accessed at http://www2.ljmu.ac.uk/corporatecommunications/60486.htm;
- The study consent forms, data, information etc. will be accessible on request to a student's supervisory team and/or to responsible members of Liverpool John Moores University for monitoring, auditing and data authenticity purposes.

Where any substantive amendments are proposed to the protocol or study procedures further ethical approval must be sought (https://www2.ljmu.ac.uk/RGSO/93205.htm)

Applicants should note that where relevant appropriate gatekeeper / management permission must be obtained prior to the study commencing at the study site concerned.

Please note that ethical approval is given for a period of five years from the date granted (23/05/19) and therefore the expiry date for this project will be 5 years from the approval date. An application for extension of approval must be submitted if the project continues after this date.

23rd December 2019
With reference to your Application for Ethical Approval
Carreon, Richie-Paul PGR - The voices of Huntington's Disease (HD)

UREC decision: Approved with provisos
UREC reference: 19/NAH/040

The University Research Ethics Committee (UREC) has considered the above application. I am pleased to inform you that ethical approval has been granted subject to the provisos listed

below. Once the final version of the ethics application with the provisos addressed has been emailed to FullReviewUREC@ljmu.ac.uk, the study can commence.

(Please note, UREC will not check that the provisos have been applied in the final version of the ethics application and will not email any further approval notifications to the applicant once the final version of the ethics application has been forwarded to UREC. If the applicant does not want to apply the provisos as stated below, the applicant must notify UREC and resubmit the ethics application for further review)

Provisos:

1. Start date might need moving back to Jan 2020, or at least late Dec 2019
2. F – make clear contact information will not be stored with data.
3. F – Please explain how case study data be coded and linked
4. PIS – Please include more information for wider involvement e.g. family members, healthcare professionals - to inform participant of the wider case study work

Approval is given on the understanding that:

- any adverse reactions/events which take place during the course of the project are reported to the Committee immediately by emailing FullReviewUREC@ljmu.ac.uk
  any unforeseen ethical issues arising during the course of the project will be reported to the Committee immediately emailing FullReviewUREC@ljmu.ac.uk
- the LJMU logo is used for all documentation relating to participant recruitment and participation e.g. poster, information sheets, consent forms, questionnaires. The study consent forms, data, information etc. will be accessible on request to a student's supervisory team and/or to responsible members of Liverpool John Moores University for monitoring, auditing and data authenticity purposes.

Where any substantive amendments are proposed to the protocol or study procedures further ethical approval must be sought (https://www.ljmu.ac.uk/ris/research-ethics-and-governance/research-ethics/university-research-ethics-committee-urec/amendments)

Applicants should note that where relevant appropriate gatekeeper / management permission must be obtained prior to the study commencing at the study site concerned.

Please note that ethical approval is given for a period of five years from 18/12/2019 and therefore the expiry date for this project will be 5 years from the approval date. An application for extension of approval must be submitted if the project continues after this date.

10th October 2021

The voices of Huntington's Disease.

UREC reference: 19/NAH/040

Research Ethics and Governance

UREC opinion: Favourable ethical opinion with provisos

Research Governance Assessment: Approved – the amended study may commence

On behalf of the University Research Ethics Committee I am pleased to confirm a favourable ethical opinion of the amendment on the basis described in the Study Amendment Form, supporting documents and any clarifications received, subject to the provisos and conditions specified below. You are required to email the final version of the amendment form with the provisos addressed to FullReviewUREC@ljmu.ac.uk. Please note, UREC will not check that the provisos have been applied in the final version of the amendment form and will not email any further notifications to the applicant once the final version of the amendment form has been forwarded to UREC. If the applicant does not want to apply the provisos as stated below, the applicant must notify UREC and resubmit the amendment form for further review.

Provisos:

1. Face-to-face meetings with participant will only take place if the investigator has obtained a negative lateral flow test immediately prior to the meeting.
2. Face-to-face meetings will only take place inside if it is not possible to meet outside or if not possible to rearrange to meet outside at another time.

Conditions of the favourable opinion

Prior to the start of the study.

- Covid-19. Studies that involve face-to-face activity – you must ensure participant facing documents explain the potential risks of participating in the study which are associated with Covid-19, how the risks will be mitigated and managed.

After ethical review.

- You must ensure the information included in the participant facing documents are always current and informed by ongoing risk assessments and any changes to current practices.
- Where any substantive amendments are proposed to the protocol or study procedures further ethical opinion must be sought (https://www.ljmu.ac.uk/ris/research-ethics-and-governance/research-ethics/university-research-ethics-committee-urec/amendments)
- Any adverse reactions/events which take place during the course of the project are reported to the Committee immediately by emailing FullReviewUREC@ljmu.ac.uk
- Any unforeseen ethical issues arising during the course of the project will be reported to the Committee immediately emailing FullReviewUREC@ljmu.ac.uk

Research Governance Approval.

This email also constitutes LJMU Research Governance Approval of the amended study, on the basis described in the Study Amendment Form, supporting documents and any clarifications received, subject to the conditions specified below.

Conditions of Approval

- Compliance with LJMU Health and Safety Codes of practice and risk assessment policy and procedures and LJMU Code of Practice for Research
- Ensure the study is covered by UMAL
- Covid-19. Compliance with LJMU's travel restrictions
- Covid-19. Studies that involve any face-to-face research activity have the appropriate risk assessment in place – the risk assessment is signed by the school Director or nominated other, revised, resigned and reissued when required and sent to the Safety, Health and Environment Department by email to SHE@ljmu.ac.uk
- Covid-19. Studies that involve any face-to-face research activity meet Covid-19 practices which are current at the time the research activity takes place.
- Where relevant, appropriate gatekeeper / management permission is obtained at the study site concerned and any other approvals that are required are obtained.
- The LJMU logo is used for all documentation relating to participant recruitment and participation e.g. poster, information sheets, consent forms, questionnaires.
- The study consent forms, study data/information, all documents related to the study etc. will be accessible on request to a student's supervisory team and/or to responsible members of Liverpool John Moores University for monitoring, auditing and data authenticity purposes.

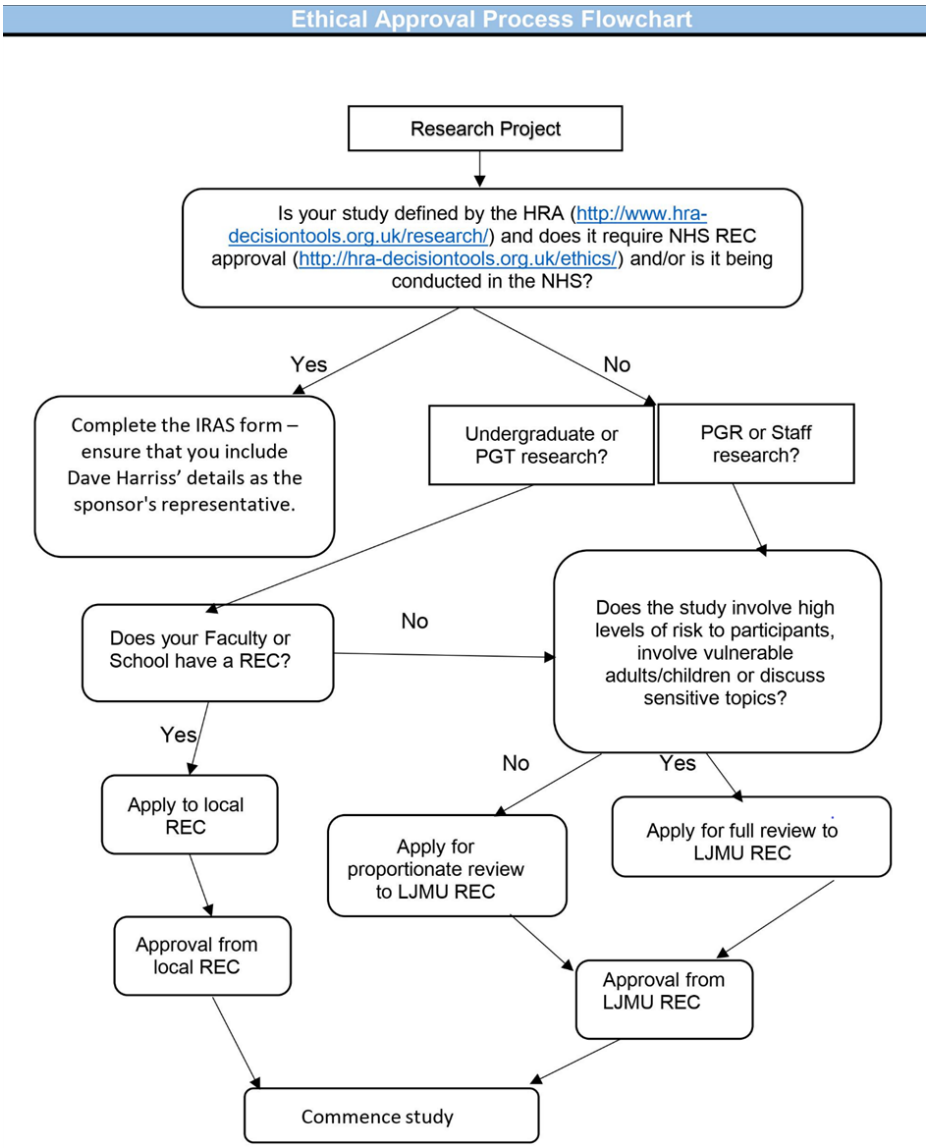


Figure 1.G. Flowchart of the approval process

Appendix H. Adapted participant information and consent forms

Information Sheet for Person with HD: Case Study

The voices of Huntington's disease (HD).

My name is **Paul Carreon.**

I am a **researcher** at **Liverpool John Moores University.**

I am a registered physiotherapist with over 15 years' experience

I have been working with people with HD for the past 8 years in the UK.

I am an active member of European HD Network Working Groups.

I would like to **invite you** to take part in a **case study research, together with your caregiver, family member or health professional.**

The case study hopes to answer the **main research question:**

1. What are the **unique needs and experiences of people with HD** in the **different stages of the disease?**

Please **take time** to read this booklet.

Please **ask questions** before you decide to participate.

Please **discuss with others,** if you have any concerns.

What is the purpose of the case study?

To try to **understand the living experiences** of people with Huntington's disease, in the different stages of the disease.

The study aims **raise awareness** for the people living with HD, share their stories which includes presenting this to conferences (UK and Abroad) and writing in research publications.

Why have I been chosen?

You have been chosen because you have Huntington's disease, and from your contact with the Huntington's disease Association.

Do I have to take part?

No. This is **voluntary.**

What does the Case Study involve?

Talking to you about your experiences of living with HD,

With your permission, I will observe you with your caregiver.

We can talk about anything memorable or personal to you relating to HD (PHOTOGRAPHS, JOURNALS, OTHERS)

I will write down notes, use the audio recorder to record our interview.

This will only take an average of one hour, three meetings in approximately 6 months.

Please remember,

You can stop at any time.

You can **start again at any time.**

You choose the day and time you want me to interview and see you.

And if you **feel upset** about talking about your condition,

I will make sure **someone is there to support you.**

What are the possible benefits of this Case Study?

This may **help us understand more about Huntington's disease**, your **experiences and challenges** on a day to day.

This may **also help people in the future** to provide **better care and understanding for Huntington's disease.**

How about my privacy and confidentiality?

You will NOT be named in the study.

Anything I hear or see will be **kept in confidential.**

I will NOT share any personal information to people outside the research team.

What will happen after the Case Study?

The study will be **written up in a PhD thesis and other Research publications.**

There will be **no personal information about you** in the study.

Who has reviewed and approved the study?

This study has been granted with **Ethical Approval** from Liverpool John Moores University, University Research Ethics Committee.

Also, advice has been obtained from the **Huntington's Disease Association and European HD Network Members.**

Where can I get more information?

Please ask me if you have any questions- **Paul Carreon**

Or I can be contacted at

Telephone: 0151 231 8737

Email: R.P.Carreon@2018.ljmu.ac.uk

Address: School of Nursing and Allied Health

Henry Cotton Building, 15-21 Webster Street, Liverpool L3 2ET

Thank you.

LIVERPOOL JOHN MOORES UNIVERSITY

HD Participant Consent Form

Case Study

The voices of Huntington's disease (HD).

I have seen the Case Study Information sheet

YES	NO
☐	☐

I have spoken to either my relative, carer or someone involved in my care about the Case Study

YES ☐ 

NO ☐ 

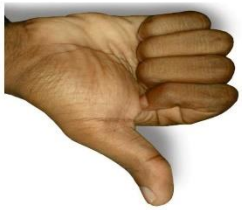
We have looked at the information sheet together

YES ☐ 

NO ☐ 

I have talked to Paul Carreon

YES **NO**



My questions have been answered

YES **NO**

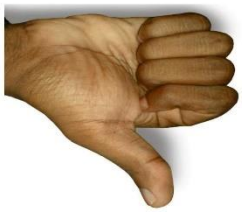
I have understand my involvement in the Case Study

YES **NO**

I understand it is voluntary and my free choice,

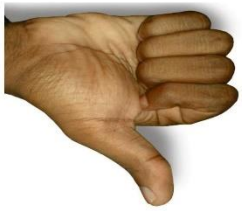
I can stop anytime.

YES **NO**



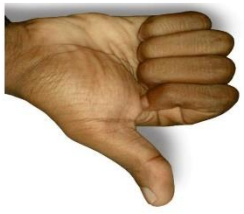
I agree to participate in the Case Study Research

YES ☐ 

NO ☐ 

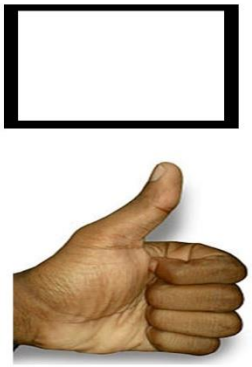
I agree that interviews will be audio-recorded.

YES  ☐ 

NO ☐ 

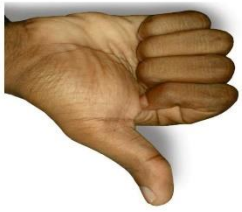
I agree that Capacity Assessments will be regularly taken in the study.

YES **NO**

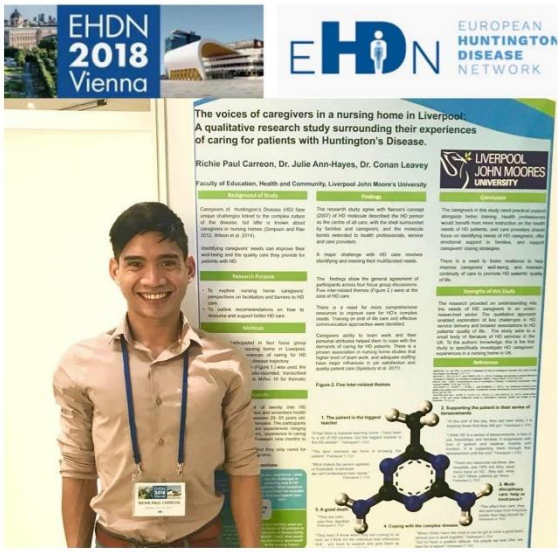


I agree this information will be used in Paul's PhD research,

YES **NO**



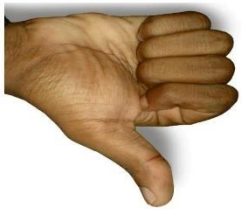
And other reports and conference publications relating to HD.



I will not be named, and my personal information will be confidential.

YES **NO**

☐ ☐



My Name: _____

Signature: _____

Date: _____

Details of person taking or witnessing consent:

Name: _____

Signature _____

Date: _____

Time: _____

Location: _____

Appendix I. Standard participant information sheets and consent forms

LIVERPOOL JOHN MOORES UNIVERSITY

Participant Information Sheet

For Person with HD (Case Study)

Research Ethics Committee Approval Reference: 19/NAH/040

YOU WILL BE GIVEN A COPY OF THIS INFORMATION SHEET

Title of Study: The voices of Huntington's disease (HD).

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

1. Who will conduct the study?

This PhD research study is under the Faculty of Health, School of Nursing and Allied Health.
The details of the researchers are:
Principal Investigator:
Richie Paul Carreon, email: R.P.Carreon@2018.ljmu.ac.uk
Supervisors:
Dr Julie Ann- Hayes, email: J.Nicholson@ljmu.ac.uk
Dr Conan Leavey, email: C.Leavey @ljmu.ac.uk
Mr Donal Deehan, email: D.Deehan@ljmu.ac.uk

2. What is the purpose of the study?

This PhD study has identified that the voice of people affected by HD is lacking in qualitative research and existing literature. The case study is a PhD led project conducted for the purpose of completing a PhD programme, and the results of this research will be used in conferences and research publications.

This case study will focus on you, the person diagnosed HD and your views to help us understand your unique day to day experiences of living with HD. The case studies will also seek to involve your families, caregivers and/or health professionals, should they wish to be involved. By combining these multiple voices of people affected by HD, this research hopes to illuminate the experiences of people affected by HD at different points in the disease trajectory. This research will provide an in-depth exploration of living with HD specific to the needs and problems of the 5 clinical stages through 5 case studies.

3. Why have I been invited to participate?

You have been invited to participate in the case study because of your experience as person diagnosed with HD.

Inclusion Criteria
• Clinical diagnosis of HD
• Age 18 and over
• Able to make informed decisions/consent

Exclusion Criteria
• Participants age 17 and under
• Participants unable to make decision informed consent
• Participants unable to retain, weigh and understand information
• Participants unable to communicate their decisions (verbal or non-verbal)

4. Do I have to take part?

Your participation is voluntary. It is up to you to decide whether or not to take part. If you do decide to take part you will be given this information sheet to keep, and be asked to sign a consent form. You can withdraw at any time by informing the researchers without giving a reason and without it affecting your rights and the service you receive.

5. What will happen to me if I take part?

The case study research will be held at your usual place of care residence or your area of preference for the interview and observations. This will take about an hour or less. You will have time for questions before and after the session.

The interviews will be audio recorded, please consider if you are comfortable with this as this is essential for your participation. I will ask some questions about your day to day experiences of living with HD. The interview session will be done in average of 3 times in the 6 months' period. The dates and times will be dependent on your availability.

The observations will be conducted on your invitation to be observed an aspect of your life. This could be anything that you would like to share to the researcher, such as your hobbies, functional tasks, aspects of your care or social events. The observations will last approx. 30 minutes and will be recorded by pen and paper.

Also, with your consent we will explore the use journals, video diaries, photographs or other artefacts to express your views on different subjects that are important to you. These are only examples, and is entirely up to you, with your choice of methods of communication that you want to use and the information you want to share. The information you will provide will only be used for analysis. No other use will be made of them without your written or recorded permission.

The research study is about capturing your experiences with HD, and you will determine how your story will be best told and shared to the researcher.

6. What are the possible benefits of taking part?

There will be no direct benefits to you, but this study hopes that this PhD will be able to contribute to the field of qualitative health research, by raising awareness and prioritising research agenda through the voices of people affected by HD.

7. Are there any possible disadvantages or risks from taking part?

The case study will include discussions on your experiences of living with HD. Some participants might find these topics sensitive or distressing. If you are personally affected by participation in this study, you may wish to seek support and advice from Huntington's disease Association Specialist Advisor and the local HD support group. Telephone: 0151 487 6514.

8. What will happen to the data provided and how will my taking part in this project be kept confidential?

The information you provide as part of the study is the study data. Any study data from which you can be identified (e.g. from identifiers such as your name, date of birth, audio recording etc.), is known as personal data. This includes more sensitive categories of personal data (sensitive data) such as your race; ethnic origin; politics; religion; trade union membership; genetics; biometrics (where used for ID purposes); health; sex life; or sexual orientation. Personal data collected from you will be recorded using a linked code – the link from the code to your identity will be stored securely and separately from the coded data

You will not be identifiable in any ensuing reports or publications. We will use pseudonyms in transcripts and reports to help protect the identity of individuals and organisations unless you tell us that you would like to be attributed to information/direct quote etc.

When you agree to take part in a study, we will use your personal data in the ways needed to conduct and analyse the study and if necessary, to verify and defend, when required, the process and outcomes of the study. Personal data will be accessible only to the study team and transcription service. The study team includes the researcher's supervisors.
When we do not need to use personal data, it will be deleted or identifiers will be removed. Personal data does not include data that cannot be identified to an individual (e.g. data collected anonymously or where identifiers have been removed). However, your consent form, contact details, audio recordings etc. will be retained for 5 years.

9. What will happen to the results of the study?

The principal researcher intends to publish the results in a PhD thesis and the research to be disseminated in the conference presentations and journal publications.

10. Who organised and reviewed this study?
This study is organised by Liverpool John Moores University, looking at the lived experiences of people diagnosed and affected by HD. The organisation and the research team declares no conflict of interest in this study.

This study has been reviewed by, and received ethics clearance through, the Liverpool John Moores University Research Ethics Committee (Reference number:19/NAH/040).

11. What if something goes wrong?
If you have a concern about any aspect of this study, please contact the relevant investigator who will do their best to answer your query. The investigator should acknowledge your concern within 10 working days and give you an indication of how they intend to deal with it. If you wish to make a complaint, please contact the chair of the Liverpool John Moores University Research Ethics Committee (researchethics@ljmu.ac.uk) and your communication will be re-directed to an independent person as appropriate.

12. Data Protection Notice

Liverpool John Moores University is the sponsor for this study based in the United Kingdom. We will be using information from you in order to undertake this study and will act as the data controller for this study. This means that we are responsible for looking after your information and using it properly. Liverpool John Moores University will process your personal data for the purpose of research. Research is a task that we perform in the public interest. Liverpool John Moores University will keep identifiable information about you for 5 years after the study has finished.

Your rights to access, change or move your information are limited, as we need to manage your information in specific ways in order for the study to be reliable and accurate. If you withdraw from the study, we will keep the information about you that we have already obtained. To safeguard your rights, we will use the minimum personally-identifiable information possible.

You can find out more about how we use your information at URL and/or by contacting secretariat@ljmu.ac.uk.

If you are concerned about how your personal data is being processed, please contact LJMU in the first instance at secretariat@ljmu.ac.uk. If you remain unsatisfied, you may wish to contact the Information Commissioner's Office (ICO). Contact details, and details of data subject rights, are available on the ICO website at: https://ico.org.uk/for-organisations/data-protection-reform/overview-of-the-gdpr/individuals-rights/

13. Contact for further information

Richie Paul Carreon, BSc Hons Phys, PgCert CE, MSc IPH
PhD Researcher
Email: R.P.Carreon@2018.ljmu.ac.uk,
Tel. No. 01512318737

Liverpool John Moores University
School of Nursing and Allied Health
Faculty of Health
Henry Cotton Building, 15-21 Webster Street,
Liverpool L3 2ET

Thank you for reading this information sheet and for considering to take part in this study.

Note: A copy of the participant information sheet should be retained by the participant with a copy of the signed consent form.

LIVERPOOL JOHN MOORES UNIVERSITY

CONSENT FORM

The voices of Huntington's disease (HD).

1. **I confirm that I have read and understand the information provided for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily** ☐
2. **I understand that my participation is voluntary and that I am free to withdraw at any time, without giving a reason and that this will not affect my legal rights.** ☐
3. **I understand that any personal information collected during the study will be anonymised and remain confidential** ☐
4. **I agree to take part in the Case Study Research** ☐
5. **I understand that the interview will be audio recorded and I am happy to proceed** ☐
6. **I agree to take part with the participant observations** ☐
7. **I understand that parts of our conversation may be used verbatim in future publications or presentations but that such quotes will be anonymised.** ☐
8. **I understand that by taking part in the Case Study, I will be indirectly identifiable due to small study sample. The researcher will work with me in an attempt to minimise and manage the potential for indirect identification of participants.** ☐

Name of Participant: _____

Date: _____

Signature: _____

Name of Person taking/witnessing Consent:

_____

Date: _____

Signature: _____

Name of Researcher:

Richie Paul Carreon,

Faculty of Health

School of Nursing and Allied Health

Date :

Signature:

Note: When completed 1 copy for participant and 1 copy for researcher

LIVERPOOL JOHN MOORES UNIVERSITY

Participant Information Sheet

For HD Families, Carers and Health Professionals

LJMU's Research Ethics Committee Approval Reference: 19/NAH/040

YOU WILL BE GIVEN A COPY OF THIS INFORMATION SHEET

Title of Study: The voices of Huntington's disease (HD).

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

1. Who will conduct the study?

This PhD research study is under the Faculty of Health, Education and Community, School of Nursing and Allied Health.
The details of the researchers are:

Principal Investigator:
Richie Paul Carreon, email: R.P.Carreon@2018.ljmu.ac.uk
Supervisors:
Dr Julie Ann- Hayes, email: J.Nicholson@ljmu.ac.uk
Dr Conan Leavey, email: C.Leavey @ljmu.ac.uk
Donal Deehan, email: D.Deehan@ljmu.ac.uk

2. What is the purpose of the study?

This PhD study has identified that the voice of people affected by HD specifically in advanced stages of the disease is lacking in qualitative research and existing literature. Further, the views of families, carers and health professionals involve in HD care will illuminate a holistic picture of the living experience of HD, and the experiences of people living.

The case study is a PhD student led project conducted for the purpose of completing a PhD programme and the results of this research will be used in conferences and research publications.

This case study will focus on the person with HD as the principal component, and your views as a person involved with the care of people with HD will hope to help us understand the unique living experiences of HD in different trajectories.

3. Why have I been invited to participate?

You have been invited to participate in the case study because of your experience as a family member, a caregiver or a health professional caring for someone with HD.

4. Do I have to take part?

Your participation is voluntary. It is up to you to decide whether or not to take part. If you do decide to take part you will be given this information sheet to keep, and be asked

to sign a consent form. You can withdraw at any time by informing the researchers without giving a reason and without it affecting your rights and the service you receive.

5. What will happen to me if I take part?

The case study research will be held at the place of care for the person with HD in the community, usually your area of preference for the interview.

I would suggest that interviews will be held at a private identified space, to maintain data privacy and confidentiality. This will take about an hour or less for interviews or observations facilitated by the principal researcher. You will have time for questions before and after the session. Information and support services from the HDA will be available, should you require it. The interviews will be audio recorded, please consider if you are comfortable with this as this is essential for your participation. The recordings made will only be used for analysis. No other use will be made of them without your written permission.

6. What are the possible benefits of taking part?

There will be no direct benefits to you, but this study hopes that this PhD will be able to contribute to the field of qualitative health research, by raising awareness and prioritising research agenda through the voices of people affected by HD.

7. What will happen to the data provided and how will my taking part in this project be kept confidential?

The information you provide as part of the study is the study data. Any study data from which you can be identified (e.g. from identifiers such as your name, date of birth, audio recording etc.), is known as personal data. This includes more sensitive categories of personal data (sensitive data) such as your race; ethnic origin; politics; religion; trade union membership; genetics; biometrics (where used for ID purposes); health; sex life; or sexual orientation. Personal data collected from you will be recorded using a linked code – the link from the code to your identity will be stored securely and separately from the coded data

You will not be identifiable in any ensuing reports or publications. We will use pseudonyms in transcripts and reports to help protect the identity of individuals and organisations unless you tell us that you would like to be attributed to information/direct quotes etc.

When you agree to take part in a study, we will use your personal data in the ways needed to conduct and analyse the study and if necessary, to verify and defend, when required, the process and outcomes of the study. Personal data will be accessible only to the study team and transcription service. The study team includes the researcher's supervisors.
When we do not need to use personal data, it will be deleted or identifiers will be removed. Personal data does not include data that cannot be identified to an individual (e.g. data collected anonymously or where identifiers have been removed). However, your consent form, contact details, audio recordings etc. will be retained for 5 years.

8. What will happen to the results of the study?

The principal researcher intends to publish the results in a PhD thesis and the research to be disseminated in the conference presentations and journal publications.

9. Who organised and reviewed this study?

This study is organised by Liverpool John Moores University, looking at the lived experiences of people diagnosed and affected by HD. The organisation and the research team declares no conflict of interest in this study.

This study has been reviewed by, and received ethics clearance through, the Liverpool John Moores University Research Ethics Committee Reference number: 19/NAH/040

10. What if something goes wrong?

If you have a concern about any aspect of this study, please contact the relevant investigator who will do their best to answer your query. The investigator should acknowledge your concern within 10 working days and give you an indication of how they intend to deal with it. If you wish to make a complaint, please contact the chair of the Liverpool John Moores University Research Ethics Committee (researchethics@ljmu.ac.uk) and your communication will be re-directed to an independent person as appropriate.

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Your rights to access, change or move your information are limited, as we need to manage your information in specific ways in order for the study to be reliable and accurate. If you withdraw from the study, we will keep the information about you that we have already obtained. To safeguard your rights, we will use the minimum personally-identifiable information possible.

You can find out more about how we use your information at URL and/or by contacting secretariat@ljmu.ac.uk.

If you are concerned about how your personal data is being processed, please contact LJMU in the first instance at secretariat@ljmu.ac.uk. If you remain unsatisfied, you may wish to contact the Information Commissioner's Office (ICO). Contact details, and details of data subject rights, are available on the ICO website at: https://ico.org.uk/for-organisations/data-protection-reform/overview-of-the-gdpr/individuals-rights/

12. Contact for further information

Richie Paul Carreon, BSc Hons Phys, PgCert CE, MSc IPH
PhD Researcher/Research Assistant
Email: R.P.Carreon@2018.ljmu.ac.uk,
Tel. No. 01512318737

Liverpool John Moores University
School of Nursing and Allied Health
Faculty of Education, Health and Community
Henry Cotton Building, 15-21 Webster Street,
Liverpool L3 2ET

Thank you for reading this information sheet and for considering to take part in this study.

Note: A copy of the participant information sheet should be retained by the participant with a copy of the signed consent form.

LIVERPOOL JOHN MOORES UNIVERSITY

CONSENT FORM

The voices of Huntington's disease (HD)

1. I confirm that I have read and understand the information provided for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily ☐
2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving a reason and that this will not affect my legal rights. ☐
3. I understand that any personal information collected during the study will be anonymised and remain confidential ☐
4. I agree to take part in the Case Study Research ☐
5. I agree to take part with the participant observations ☐
6. I understand that the interview will be audio recorded and I am happy to proceed ☐
7. I understand that parts of our conversation may be used verbatim in future publications or presentations but that such quotes will be anonymised. The researcher will work with me in an attempt to minimise and manage the potential for indirect identification of participants. ☐

Name of Participant:	Date	Signature
_____	_____	_____

Name of Researcher:	Date	Signature
Richie Paul Carreon, Faculty of Health School of Nursing and Allied Health		

Note: When completed 1 copy for participant and 1 copy for researcher

Appendix J. Mental Capacity Assessment (MCA) and study protocol

Principal Investigator's (PI) Note:

Given that the HD participants in their late stages might have an impairment of, or disturbance to the functioning of the mind or the brain, the PI will assess the person's ability to make the decision in participating in the study. In the time periods before any research procedure (interview, observation), the PI could presume the HD person has capacity, and will ask questions that would evidence their understanding on participating in the study, that they retain the information provided to them, and they can weigh the pros and cons of their decisions.

This in accordance to Mental Capacity Act 2005 **(https://www.hra.nhs.uk/planning-and-improving-research/policies-standards-legislation/mental-capacity-act/).**

Furthermore, the PI will utilise HD information and consent forms (Appendix VI) drawing from relevant literature and expert opinions which falls in line with the recommendations from the Mental Capacity Act 2005, to encourage people to make their own decisions, and the importance of on-going process of consent for people with HD (Wilson et al 2010). This PhD study places particular emphasis on adapting different forms of communication and the importance of written information designed to meet the tailored needs of people with HD in different stages of disease progression.

Mental Capacity Assessment

- A person must be assumed to have capacity unless it is established that he lacks capacity.
- A person is not to be treated as unable to make a decision unless all practicable steps to help him to do so have been taken without success.
- A person is not to be treated as unable to make a decision merely because he makes an unwise decision.
- An act done, or decision made, under this Act for or on behalf of a person who lacks capacity must be done, or made, in his best interests.
- Before the act is done, or the decision is made, regard must be had to whether the purpose for which it is needed can be as effectively achieved in a way that is less restrictive of the person's rights and freedom of action.

Mental Capacity Act 2005 https://www.gov.uk/government/collections/mental-capacity-act-making-decisions#mental-capacity-act-code-of-practice

Name:

Date of assessment:
Location of assessment:

Question1 : Diagnostic Test

Is there an impairment of or disturbance in the functioning of the patients mind or brain?
□ Yes

Detail:

□Permanent Impairment □ Temporary Impairment □ Fluctuating Impairment
□ No If there is no disturbance, there is no reason to continue the capacity assessment

What decision needs to be taken? (mental capacity assessment is decision and time specific)

Question 2: Functional Test

Is the person is able to understand the information relevant to the decision being made. Have steps been taken to maximise understanding (E.G. Easy Read / Pictures / Aids)?	□ YES □ NO Record views / evidence to show they understand it.
Can the person retain the information long enough to make a decision?	□ YES □ NO Record views / evidence to show they understand it.

Can the person weigh up the information as part of the decision making process?	□ YES □ NO Record views / evidence to show they understand it.
Can the person communicate their decision by any means?	□ YES □NO Record views / evidence to show they understand it.

FLUCTUATING CAPACITY – Always consider whether the person has fluctuating capacity and if the decision can wait until capacity returns. If this is the case, explain and enter reassessment date in the outcome below.	
CONCLUSION: If the answer to Question 1 is Yes and the answer to any parts of Question 2 is NO then the person being assessed LACKS capacity under the Mental Capacity Act 2005.	
OUTCOME:	□ Has capacity
	□ Lack capacity at this time of interaction
Assessor name: Assessor signature: Designation: Date:	
Other comments or considerations:	

Appendix K. Recruitment posters

Calling for Research Participants in HD Case Studies

People with HD in different clinical HD stages

Research Title: The voices of Huntington's disease (HD)

Introduction:

Thank you for your interest in the research study. My name is Paul Carreon, a PhD researcher at Liverpool John Moores University, and I am conducting this research for my thesis under the supervision of Dr Julie Ann Hayes, Dr Conan Leavey and Mr. Donal Deehan.

This research is promoted via the Huntington's disease Association UK, and has received LJMU Research Ethical Approval (UREC NO. 19NAH40)

About the Research:

The existing body of literature in HD features a lack of research on the lived experiences, the problems and needs of people living with HD. This PhD study aims to give voice to people living with, and affected by HD. It is with the hope to share this information to other researchers and the wider health and social community, to further increase our understanding with HD lived experiences and develop good care practices.

The case study research will involve in-depth interviews, observations and talk about other artefacts which is person-tailored to you (photographs, journals, etc.) or any methods of sharing your experiences of living with HD. The interviews also hope to include people significant in your life such as your family member, partner, caregiver or health professional, should they wish to get involve.

The interviews will last approximately an hour or less, conducted average three times in a period of approximately 6 months. If you choose to participate, this is an opportunity to share your story and provide insight to the principal case study question: *What are the unique needs of people with HD in the different stages of the disease?*

Contact:

If you are interested, please contact the details below and I will give you more information about the research, and the participant information sheets.
Thank you very much.

LIVERPOOL JOHN MOORES UNIVERSITY

Paul Carreon
Email: R.P.Carreon@2018.ljmu.ac.uk
Tel. 0151 231 8737

Person with HD
Inclusion criteria: • Clinical diagnosis of HD. • Age 18 and over. • Able to make informed decisions/consent with the study intervention.
Exclusion criteria: • Participants age 17 and under • Participants unable to make decision informed consent. • Participants unable to retain, weigh and understand information. • Participants unable to communicate their decisions (verbal or non-verbal).

Paul Carreon
School of Nursing and Allied Health
Faculty of Health
Email: R.P.Carreon@2018.ljmu.ac.uk
Tel. 0151 231 8737

LIVERPOOL JOHN MOORES UNIVERSITY

Social Media (Facebook group /Twitter) message

Hello_____,

My name is Paul Carreon, a PhD researcher at Liverpool John Moore's University.

I am currently doing a research on the experiences of people living with HD throughout the different stages of the disease progression. If you would like to more information about participating in the case study, please see the link *{inset link}* which will provide participant information sheet and information about the study.

Please feel free to contact me if you would like to know more about the research.

Thank you very much.

Paul Carreon
Email: R.P.Carreon@2018.ljmu.ac.uk
Tel. 0151 231 8737

My name is Paul Carreon, a PhD researcher at Liverpool John Moore's University.

I am currently doing a research on the experiences of people living with HD throughout the different stages of the disease progression, and I am looking for participants who would like to talk about their experiences in living with, and affected by HD.

If you would like to more information about participating in the case study, please see the link https://voicesofhuntingtons.wordpress.com/

which will provide participant information sheet and details about the study.

Please feel free to contact me via web page or phone at 0151 231 8737,

if you would like to know more about the research.

Thank you very much.

Appendix L. Acts of interpretation

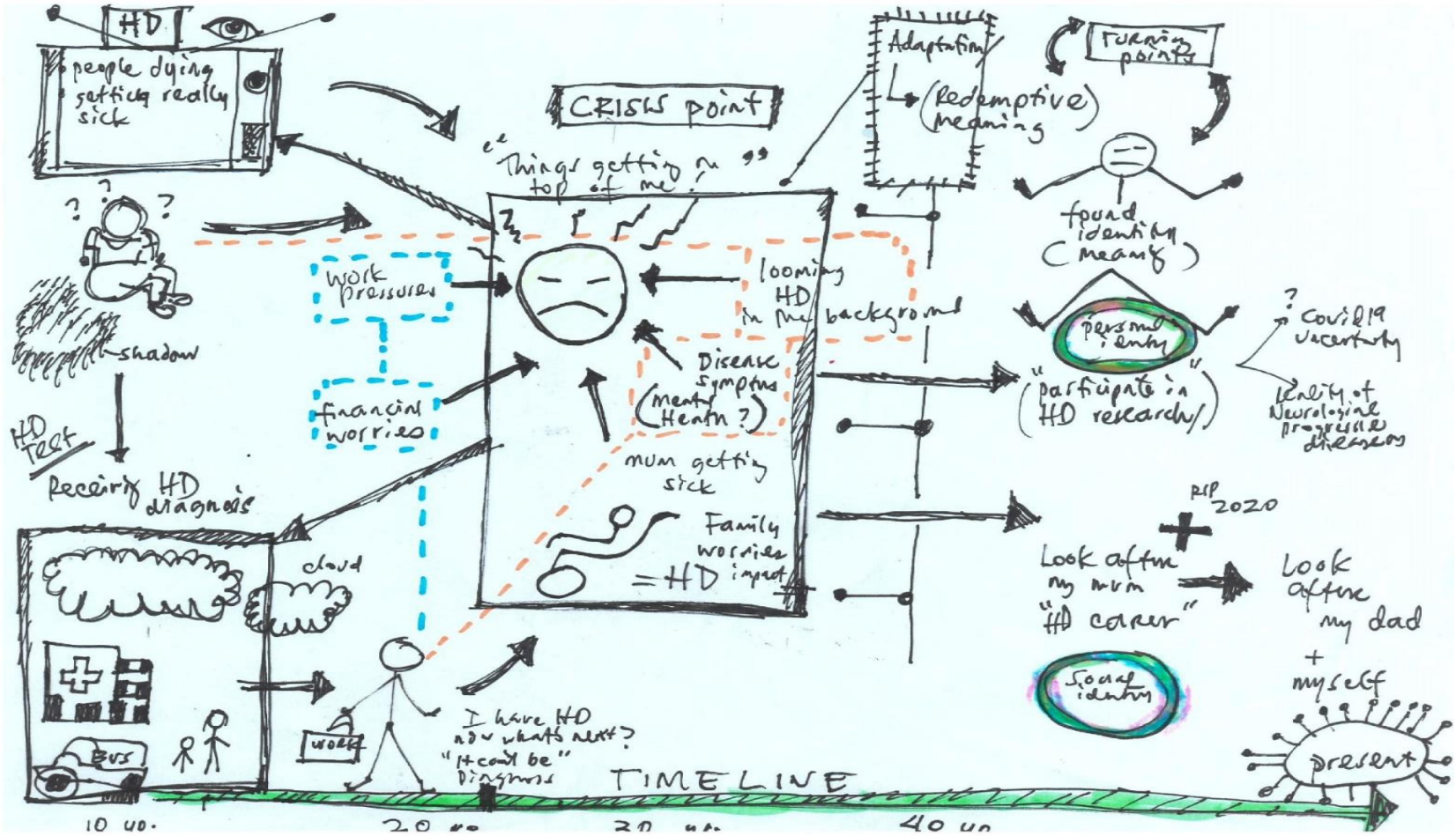


Figure 1.L Example of translating narrative to images

Summary of Martin's Narrative

This story is about man of retiring age, who suffered mental health issues over the past 10 years. That his marriage is at it breaking point following years of depression and anxiety, and his wife who had suffered verbal aggression and neglect. The paradox of while he was trying to help other people in the community as a mental health nurse, deep inside he was suffering on his own and felt no one was there to help him. His mental health symptoms progressed overtime since 2007, the day he had his first breakdown at work. That marriage has reached the breaking point it was about two years ago, and that Pamela had a breakdown herself that prompted her to say that this is not the life that she wanted. Pamela has gradually witnessed e martin's deterioration and him drifting away becoming a person this aggressive and horrible a person that she didn't marry.

Pamela made a choice to stand up and didn't accept the situation, she wasn't happy with the support Martin is getting. The medications were not helping him, as he was becoming more recluse and apathetic. All the doctors were advising them is to go to these support groups and wellbeing centres. For Pamela, this was not the life that she wanted so she went to the doctors and pushed the specialist consultants. At the time Martin was just consumed by his own mental health issues felt helpless and he just left Pamela to make the decisions. Pamela at the time is having depression brought about by the turmoil of the home situation. She was seeing her doctor and has convinced her own doctor to refer Martin to another consultant. She was adamant that there was something more than just depression with Martin. When they finally saw this consultant and Martin was officially diagnosed with Huntington's disease, rather viewed as devastating news it was welcomed with a sigh of relief. Finally, the couple had an answer. The reason for Martin's troubling behaviour and that knowing it is the disease that is beyond Martin's control. The couple found a new sense of relief, that they finally knew what is wrong and that they can work together to address the issues around the impact of the illness to Martin and their life as couple.

The diagnosis has prompted Martin to search for the presence in his family. The attention turned to his mother who was previously admitted in a psychiatric institution, received ECT treatments and spent that last days of her life in a nursing home. Martin's mother has been regarded of her character and personality that 'you wouldn't mess with'. Martin recalled he had an inclination that her mother might have HD due to the gross involuntary failing movements of her extremities and had suggested this to the nursing home staff. It was recently found out the HD was in her death certificate. For the couple, their story continues as they come into terms with HD and as they both reached a retiring age. Martin is 67 and Pamela is 65. They have both devoted their life to church as they have always been. A marriage life centred around pilgrimages, where they first met. For them they had a happy marriage, were both successful in their careers, Martin as a community nurse and Pamela had café business.

However, the disease although has kept the marriage, the uncertainty of the future looms. For Martin, he is considering that he will get worse in the future and that he doesn't want to be burden to Pamela. Martin is happy to be placed to the nursing home so that Pamela can be relieved of burden. But for Pamela, she will continue to persist to become his wife and if possible, she will care for him at home until she physically can't do it anymore. The conflicting narratives of Martin passively surrendering to the disease and Pamela striving for a sense of normality and keeping the couple life intact. Pamela's coming to terms to her own role as she shifted from being a wife to a caregiver. Pamela pointed out that the traditional couple relationship has gone, that physical intimacy has been replaced by care. Pamela emphasised that this wasn't an easy transition, she found it hard when the must sleep in separate beds for safety. Pamela is having her own fears at what if she became incapable of looking after Martin because she has her own physical illness, and if that she dies first the thought of leaving Martin scares her the most. The couple both addressed the inevitable of planning for their funerals, setting money aside for it and Pamela talking to her family, to her brother about Martin's medication and routine.

For Martin he accepted that he would become worse eventually. He is coming to terms of finding his identity on what badge of illness he wants to be associated with, the depression or HD but he said the lines are blurred, for now all is good, and he doesn't want to rock the boat.

Figure 2. L Example of a Narrative Summary

Dear Daniel,

Thank you for sharing with me your life story. I felt privileged to hear your journey with your illness, not only on what it is like to live with HD but also how to care for your mum until the very end. Your stories had helped me understand the unique perspective of the dual nature of your condition, how it affected you. As an HD person caring for another HD person. What struck me the most was your optimism. I think it is very admirable given through all your difficulties and troubles, you recounted and looked at life with no remorse. You embraced your fate, the future with open arms and recounted your past with longingness and great fondness.

You spoke in great lengths about your mother, your regard for her strength, character and her own way of dealing with HD. It is understandable given your recent loss that you found solace in our talks; a space to grieve. I hope my listening ear have comforted you in some way. I am most thankful for your trust. You confided in me a week after your mother's funeral, and during those difficult times you were both dealing with a personal loss and the unprecedented impact of the pandemic. Given that you haven't met me in person, and all you knew of me was my name on the research study, you bestowed me trust. Thank you for reminding me of my Ethical responsibility in this research study, that the precarious nature of my illness and its profound effect on the lives of the people I study. You reminded me that by living with this terminal illness in a pandemic, it makes death and suffering even closer. Again, thank you for opening up these conversations and realisations about these painful truths of life. What struck me the most is how you viewed your caring responsibility to your mother, not only as your responsibility but also as your vocation. How you constantly reminded me that caring for your mum was the best thing you have ever done in your life and that having HD made you a better person. Thank you for sharing those reflections, which has provided me how I profoundly viewed my own life and reminded me the virtue of being grateful for what life gives us.

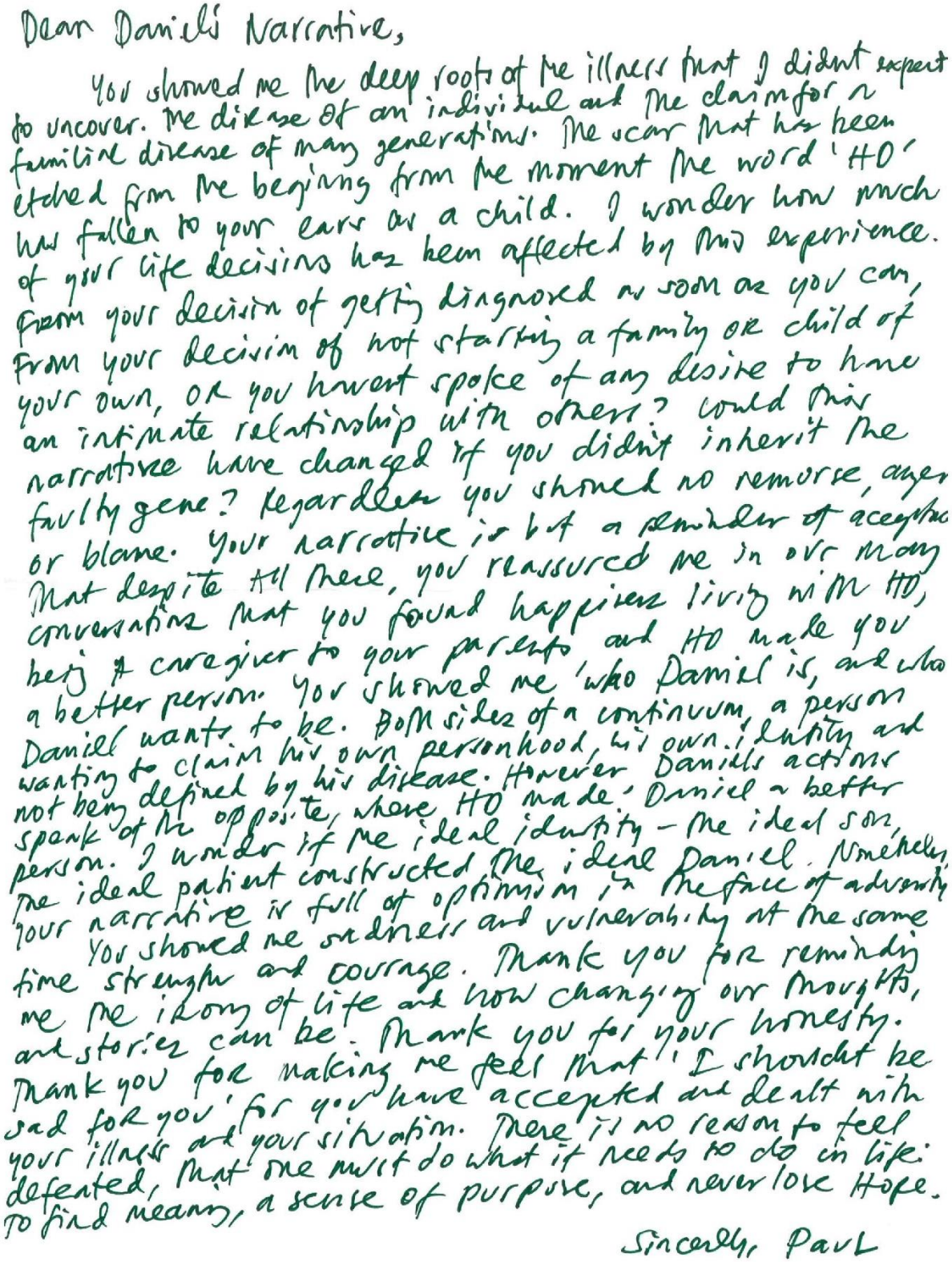
When we talked about your illness experience, you referred that having HD is not just about you, but also the story of your family. I wonder what greater impact your illness has brought to your social relationships and personal life that you devoted the rest of your life to care for your mum back back. Also, you spoke with great enthusiasm with participating with any HD related research and volunteering to HD events. This make me think, that you focused more on what you can do, rather than the things that you can't do nothing about. Thank you for reminding me this important lesson that perhaps all we need to focus on life are the things we can control and have the serenity to accept the things that we couldn't change.

Finally, your stories portray that in the face of living with the inevitable progression of your illness, you showed no fear of what is to come. You showed your own way to accept your illness and chose to be defined by your actions rather than the disease. You placed importance on the role of families and being there for them. Thank you for reminding me that in the end, our Family and love ones are the real treasures we have in life and when everything goes, they are the only one to stay behind or beside us. Thank you for sharing your life lessons. And Despite you feel that the cure for HD is far beyond your lifetime, you continue to live your life everyday that there is still something worth doing or living for. As you aptly put it, "we all live in Hope!"

Sincerely Yours,

Paul

Figure 3.L Example letter to the participant


Dear Daniel's Narrative,

You showed me the deep roots of the illness that I didn't expect to uncover. The disease of an individual and the claim for a familial disease of many generations. The scar that has been etched from the beginning from the moment the word 'HD' has fallen to your ears as a child. I wonder how much of your life decisions has been affected by this experience. From your decision of getting diagnosed as soon as you can, from your decision of not starting a family or child of your own, or you haven't spoke of any desire to have an intimate relationship with others? Would this narrative have changed if you didn't inherit the faulty gene? Regardless you showed no remorse, anger or blame. Your narrative is but a reminder of acceptance that despite all these, you reassured me in our many conversations that you found happiness living with HD, being a caregiver to your parents and HD made you a better person. You showed me 'who Daniel is, and who Daniel wants to be. Both sides of a continuum, a person wanting to claim his own personhood, his own identity and not being defined by his disease. However Daniel's actions speak of the opposite, where HD made Daniel a better person. I wonder if the ideal identity - the ideal son, the ideal patient constructed the ideal Daniel. Nonetheless your narrative is full of optimism in the face of adversity.

You showed me sadness and vulnerability at the same time strength and courage. Thank you for reminding me the irony of life and how changing our thoughts, and stories can be. Thank you for your honesty. Thank you for making me feel that I shouldn't be sad for you, for you have accepted and dealt with your illness and your situation. There is no reason to feel defeated, that one must do what it needs to do in life. To find meaning, a sense of purpose, and never lose Hope.

Sincerely, Paul

Figure 4.L Example of a letter to the narrative

Appendix M. Covid-19 risk assessments

LIVERPOOL JOHN MOORES UNIVERSITY

INDIVIDUAL RISK PROFILE: R.P.Carreon, PHD Student

The purpose of this form is to assist staff in identifying personal level of risk and to ensure any risks are identified and appropriate measures put in place to minimise and manage those risks.

DATA PROTECTION

All information provided within this document will remain confidential and secure in line with the General Data Protection Regulation and will only be used for the purpose of the COVID-19 individual risk self-assessment. Any recipient of an individual's self-assessment must not share any information contained without the individual's consent. If a referral to the Occupational Health Doctor is required then you will be asked to provide consent to share the information contained within this self-assessment form.

In additional, an individual can choose to select 'prefer not to say' to any of the question. However, this will impact on the overall risk score that an individual receives.

1. SHIELDING

Have you received a 'shielding letter' from the Department of Health and Social Care informing you that you are extremely clinically vulnerable?

Yes	
No	√
Prefer not to say	

If you have received a shielding letter you are automatically placed in the higher risk category - you should still consider the other factors in the self-assessment form to identify any other relevant risks.

2. SELF ASSESSMENT OF RISK FACTORS (highlight all that apply & calculate your score)

Risk Factor		Please Tick	Risk score
Age	16-49	√	0.0
	50-59		1.0
	60-69		2.0

	Prefer not to say		-
Gender assigned at birth	Male	√	1.0
	Female		0.5
	Prefer not to say		-
Health Condition	**Heart Disease** *(i.e. hypertension on treatment, post myocardial infarction, heart failure, cardiac arrhythmia treatment, heart surgery, valve disease etc.)*		1.5
	Diabetes mellitus on treatment (insulin or tablets)		1.5
	Chronic Lung Disease ***Asthma*** *needing regular steroid inhaler, recent short courses of steroid tablets, immunosuppressive drugs, current symptoms or past hospital admission* ***COPD****, fibrosing lung disease, bronchiectasis and cystic fibrosis etc who have not had shielding letter*		1.5
	Chronic **kidney disease** needing secondary care monitoring		1.5
	Obesity – BMI more than 40: use the BMI calculator tool below https://www.nhs.uk/live-well/healthy-weight/bmi-calculator/		1.0
	Immunosuppressive therapy *(steroid and other immunosuppressive medication) including HIV/AID. Please check advice given by the GP or specialist clinic on the risk to COVID*		2.0
	Recent history of **cancer** (within 1 year) or past history of **Lymphoma** or **leukaemia** in remission		2.0

	Chronic **neurological conditions that affects breathing** *(muscular dystrophy, myasthenia, Parkinson disease, MND, MS, bulbar palsy) and* ***cerebral palsy or learning difficulty***		2.9
	Sickle Cell disease (*Not trait) Thalassaemia or other blood disorders under specialist clinic (not trait)*		1.5
	Prefer not to say		-
Ethnicity	Belong to a **Black, Asian or other minority ethnic (BAME) group** (For the purposes of this document, the available evidence states that BAME groups from a primarily Black and Asian background are considered to be more at risk than other ethnic minority groups).	√	1.0
	Prefer not to say		-

Total risk score		2.0 = Low risk
Low Risk Score of 0 – 3.9	Moderate risk Score of 4 – 6.9	Higher risk Score >7 or in receipt of a shielding letter

3. PREGNANCY

Are you pregnant?	If you are pregnant please contact your HR Business Partner who will be able to provide you with further advice.

If your score puts you in the moderate or higher risk category you should arrange a meeting with your line manager as soon as possible (please note that this may be done remotely). You do not need to share your self assessment form if you do not wish to, however, it may be useful to share this form with your line manager to inform the discussion. You may also discuss the form with your HR Business Partner or Moni Akinsanya (Equality, Diversity and Inclusion Manager).

You may also wish to use this completed form as a basis for a conversation with your GP or, if relevant, your Consultant.

Your line manager will then review the local risk assessment and any medical advice and consider the measures that have already been put in place to protect staff members and make any further adjustments as appropriate. Your line manager may advise that a referral is made to the LJMU Occupational Health Doctor to obtain further advice about adjustments that can be put in place to ensure that you can return to the workplace safely.

This individual risk self-assessment has been adapted for LJMU use from the COVID-19 risk assessment produced by St Georges and circulated by the Medical Schools Council. The risk assessment is currently being used across the North West Region for healthcare students prior to participation in clinical practice. As LJMU did not develop the scoring mechanism the risk scores have not been changed.

Covid19 Risk Assessment			
Building	N/A	Date of Risk Assessment	25.05.2021
School/Service Department	Nursing and Allied Health	Assessment carried out by	Richie Paul Carreon
Location	MS Teams	Signed	rpcarreon Student No. 7139078
Activity	Discussion	Person consulted during the Risk Assessment	Supervisory Team: Julie Ann-Hayes, Conan Leavey, Donal Deehan
STEP 1 What are the Hazards?	15 months into the study, the participants have engaged in a longitudinal study on exploring the experiences of living with Huntington's Disease. In the onset of Covid19, by March 2020, the research interaction has moved to remote data collection. Majority of the data collection has been completed and in the final stages of the research process, in line with the adopted methodology, the researcher will collaborate with the participants on the research findings. The researcher and relevant participants have expressed interest to meet in a public space for a short discussion on the study, as well as a social occasion to conclude the final parts of the research. The researcher, as supported by the supervisory team discussed and unanimously agreed that in a risk assessed and safe environment, the face to face interaction is integral part of the research. This will allow the researcher to observe the changes and impact of the disease to the individual, but most importantly to maintain the positive participant-researcher relationship and an opportunity for further questions/concerns that cannot be easily communicated in remote interactions. The research interaction will not take place on LJMU premises, therefore are no perceived hazards with regards to staff or students of LJMU. There are no physical interventions in the study, there will be no physical contact with the participants. The risk of Covid 19 transmission will be mitigated by ensuring the researcher will obtain negative Rapid Antigen Test obtained within a week. Further, no researcher interaction will take place if any of the parties involved display any Covid19 related symptoms, as postulated in NHS website: https://www.nhs.uk/conditions/coronavirus-covid-19/symptoms/ The research interaction will take place in a Covid risk assessed public space, preferably in a city centre (Liverpool) café. Both parties will adhere to health and safety protocols implemented within relevant organisations and specific premises, this includes use of wearing of face masks in communal areas, hand sanitisers, socially distance chairs. If applicable and weather pending, the interaction will take place outdoors, in a garden or outdoor seating area. The interaction will adhere to social distance measures and safety precautions as outlined in the Public Health England and Government guidelines: https://www.gov.uk/government/publications/how-to-stop-the-spread-of-coronavirus-covid-19/how-to-stop-the-spread-of-coronavirus-covid-19		
STEP 2 Who might be harmed and how?	Researcher: Individual risk assessment has been completed. Low risk profile score= 2.0 Participants: Researcher have recently corresponded to case study participants. The participants who expressed interest for face to face interactions have stated that they obtained their first dose of Covid19 vaccine. Further, both parties will adhere to NHS, PHE and government guidelines to reduce the transmission of Covid19.		
STEP 3 (a) What are you already doing? *What is already in*	Researcher to wear face masks, observe good hand hygiene and social distance measures. Researcher awareness of physical environment including safe access and regress. Researcher to inform supervisors on travel plans and report (text, phone, email) before and after data collection.		

place to reduce the likelihood of harm, or to make any harm less serious	Researcher to be aware of mode of travel (train, bus, taxi) and logistics involved in field research. Researcher to ensure mobile phone is at hand with contact information readily available in the event of emergency.
STEP 3 (b) What further action is needed?	A week before the planned research interaction, the researcher will ensure a negative rapid antigen test. Consequently, the researcher will contact the participants to ensure they display no Covid19 related symptoms and in general good health. Three days before the research interaction, the researcher will contact the public premises, i.e. café. This is to ensure a reserved area will be allocated and if weather pending, to locate an outdoor seating. The day before the research interaction, the researcher will liaise with the participants and discuss adhere to safety measures as stipulated above.
STEP 4 How will you put the assessment into action?	1. Researcher to adhere to organisational health and safety policies and procedures. 2. Researcher to monitor and evaluate risks assessments. Researcher to inform supervisors, and address potential issues, when required. 3. Researcher to review risk assessments, as necessitated by changes.

LIVERPOOL JOHN MOORES UNIVERSITY

Assessment Number:
To be completed locally

COVID-19 Risk Assessment form

How to use this form	This form records you and your team's risk assessment discussions on issues and the controls needed to be put in pace to reduce the risk from the COVID-19 virus. It should sit alongside your risk assessments for other activities. It covers University, building, team and individual issues (similar to those that would be discussed with a new starter), some of which you can decide upon and some set centrally for consistency, using good practice guidance. The form follows a set structure to help you and your team – but not all of it may be applicable, so use 'Nil' or 'Not applicable' as appropriate. If you have too much detail for the main form, then record it as an 'Annex' at the end of the form, or hyperlink it electronically. Some of the form has been populated. **You must check that it applies**. Use the prompts and suggested text *shown in grey italics* – **but please make it appropriate to you and your team's tasks.** Alongside this risk assessment, it would be advisable to carry out a form of equality impact assessment - https://teams.ljmu.ac.uk/3/POD/EDI%20LJMU%20Only%20Document%20Library/guidance.pdf (if you need help contact the EDI Team- https://www.ljmu.ac.uk/staff/edi/meet-the-edi-team Following consultation with staff and their representatives, send the completed form to SHE@LJMU.ac.uk.

General information about the team, its location and activities

Building(s)	N/A	**Date of Risk Assessment**	**25.05.2021**
School/Service Department	Nursing and Allied Health	**Assessment carried out by**	Richie Paul Carreon, PhD Student
Location(s)	Liverpool city centre cafe	**Signed**	*rpcarreon*
Activities	Social distanced interaction (handing over of research documents i.e.transcripts and opportunity for questions)	**Persons consulted during completion of the Risk Assessment**	PhD Supervisors: Dr Julie Ann Hayes, Dr Conan Leavey, Mr Donal Deehan
Reason for a risk assessment specifically for COVID-19	The COVID-19 pandemic is a hazard that requires additional controls and changes in behaviour to reduce the risk of infections and ongoing transmission. The overarching hazard is: • Staff/students/visitor/contractor introduces COVID-19 infection to the premises, leading to other premises users become infected (and subsequent transfer of infection onwards). Infection transfer is by: o Droplet transfer or inhalation when in close contact o Touching contaminated surfaces and then touching their eyes, nose or mouth. **Vulnerable and clinically extremely vulnerable groups are particularly at risk, due to weakened immune response.**		

Details of staff who will be returning to work

This risk assessment covers:				
Phase 1 of the University recovery (core staff and those who cannot work from home: 28th June 2020 to end of August 2020)	Tick	Phase 2 (September 2020, academic delivery begins)	✓	Please note that a reviewed and updated risk assessment is required for Phase 2.

For this Phase, the number of team members in work is:	Number: Please include details about any rotas that may be implemented in the assessment below.	Of these, please confirm the number who are:	Number:
		Vulnerable – see COVID definition	2
		Extremely vulnerable – see COVID definition	1
		Persons with reduced mobility	
		Blind/partially sighted/ "touch" dependent	
		Other higher risk groups/categories i.e. those with PEEPS, other health issues etc.	
		Colleagues who have been provided with equipment resulting from a referral from Occupational Health	
		First Aiders	Include name(s) and days available
		Fire Warden/Evacuation Coordinator/Evacuation Chair Operators	Include name(s) and days available
All other protected characteristics as listed in the Equality Act 2010 should also be considered - https://www.ljmu.ac.uk/staff/edi/equality-act-2010 To do an Equality Impact Assessment see: https://teams.ljmu.ac.uk/3/POD/EDI%20LJMU%20Only%20Document%20Library/guidance.pdf			

This team's risk assessment also covers the following numbers of: *(zero or approximate numbers, if directly applicable)*					
Contractors		Visitors	3	Students	
Researchers	1	Other University staff		Others (please state)	

Risk assessment (specifically for COVID-19)

What are the hazards?	Who might be harmed and how?	What are you already doing to control the risks?	What further action do you need to take to control the risks?	Who needs to carry out the action?	When is the action needed by?	Done
Infection while entering/exiting building	Team members and additionally: *Researcher* Exposure to COVID-19 virus by: • Close contact with persons • Contact with contaminated surface	Line manager to familiarise themselves with: LJMU guidance Line manager to confirm: • Who is coming to work • Work times and shift patterns • Any lone worker issues • First Aider/Fire Warden/Evac Chair Operator coverage as necessary (dependent on which staff and total numbers coming into work) – either within the team or LJMU arrangements	Line manager to consult with and to communicate to team members and their representatives	Line manager	Prior to staff returning	*25.05.21*

What are the hazards?	Who might be harmed and how?	What are you already doing to control the risks?	What further action do you need to take to control the risks?	Who needs to carry out the action?	When is the action needed by?	Done
Infection while working in office/building or workspace	Team members and additionally: *Researcher* Exposure to COVID-19 virus by: • Close contact with persons	Line manager to obtain the specific Estate and Facilities Management local building arrangements and in conjunction with https://www.ljmu.ac.uk/microsites/moving-forward/information-for-staff confirm: • Building and office opening and closing up arrangements • Arrangements for entry doors and routes to work location • Arrangements for making your way around the building/office area • Use of lifts	Line Manager to consult with and to communicate to team members.	Line manager	Prior to staff returning	25.05.21

What are the hazards?	Who might be harmed and how?	What are you already doing to control the risks?	What further action do you need to take to control the risks?	Who needs to carry out the action?	When is the action needed by?	Done
	• Contact with contaminated surface	• Desk/seating arrangements to maintain social distancing • Use of meeting rooms/small offices • Working in a lab or communal workspace • Use of multi-user equipment • Hygiene arrangements • Welfare arrangements, including toilets • Kitchen area/catering arrangements • Arrangements for maximizing fresh air flow into work spaces • Lone working arrangements • First Aid arrangements • Fire evacuation arrangements • Additional arrangements for any higher risk persons (i.e. those of restricted mobility, who may need Evac Chair coverage) • Arrangements for any local specific response to incidents or emergencies				
Other hazards arising from but not due to COVID-19, from interactions with other hazards	Team members and additionally: *Not applicable*	**Remember: other health and safety risks may also be present. Think about your standard work tasks.** Items locally 'owned' by / responsibility of the team needing additional precautions for re-starting are: (please complete) Any tasks that uses PPE that may be affected/ interfered with by additional COVID precautionary measures (please complete): *Not applicable*	Line Manager and team to discuss and identify issues / tasks. Team to work up procedures and communicate. *Not applicable*	Line Manager to communicate to team members. Staff to note	Prior to staff returning to work	25.05.21

What are the hazards?	Who might be harmed and how?	What are you already doing to control the risks?	What further action do you need to take to control the risks?	Who needs to carry out the action?	When is the action needed by?	Done
Infection while interacting with others – students, contractors, delivery drivers and members of the public.	Team members and additionally: *(Nil or detail)* Exposure to COVID-19 virus by: • Close contact with persons • Contact with contaminated surface	Line Manager to obtain the specific E&FM local building arrangements and in conjunction with with LJMU guidance confirm: **Students:** *specify controls or Nil* **Contractors:** *specify controls or Nil* **Delivery drivers:** *specify controls or Nil* **Members of the public:** *specify controls or Nil*	Line Manager to consult with and to communicate to team members.	Line Manager.	Prior to staff interacting with others	*25.05.21*

Risk Assessment Review	Risk assessment to be reviewed after first week (25.05.2021), then after maximum one month 25.06.2021 *The risk assessments must be reviewed and updated after one week and then one month, or when there are significant changes, and consulted on with staff and their representatives.*

Appendix N. HD participants Total Functional Scale (TFC)

Adopted from Shoulson and Fahn (1979)

Name, HD Stage	**Jen Walters, Late**	
TFC Domain	Level of Functioning	Score
Occupation	Normal	3
	Reduced capacity for usual job	2
	Marginal work only	1
	Unable	**0**
Finances	Normal	3
	Slight Assistance	2
	Major Assistance	1
	Unable	**0**
Domestic Chores	Normal	2
	Impaired	**1**
	Unable	**0**
Activities of Daily Living	Normal	3
	Minimal Impairment	2
	Gross tasks only	**1**
	Total care	0

Care Setting	Home	2
	Home with chronic care	1
	Full time skilled nursing care	**0**
Total TFC Score	**2 (Start of Study)**	
	1 (End of Study)	

Table 1. N. Jen Walters Self-Reported TFC

Name, HD Stage	**Martin Taylor, Middle**	
TFC Domain	Level of Functioning	Score
Occupation	Normal	3
	Reduced capacity for usual job	2
	Marginal work only	1
	Unable	**0**
Finances	Normal	3
	Slight Assistance	**2**
	Major Assistance	**1**
	Unable	0
Domestic Chores	Normal	**2**
	Impaired	**1**
	Unable	0

Activities of Daily Living	Normal	3
	Minimal Impairment	**2**
	Gross tasks only	**1**
	Total care	0
Care Setting	Home	**2**
	Home with chronic care	1
	Full time skilled nursing care	0
Total TFC Score	**7 (Start of Study)**	
	5 (End of Study)	

Table 2. N. Martin Taylor Self-Reported TFC

Name, HD Stage	**Daniel Carragher, Early**	
TFC Domain	Level of Functioning	Score
Occupation	Normal	3
	Reduced capacity for usual job	**2**
	Marginal work only	1
	Unable	0
Finances	Normal	**3**
	Slight Assistance	2
	Major Assistance	1

	Unable	0
Domestic Chores	Normal	**2**
	Impaired	1
	Unable	0
Activities of Daily Living	Normal	**3**
	Minimal Impairment	2
	Gross tasks only	1
	Total care	0
Care Setting	Home	**2**
	Home with chronic care	1
	Full time skilled nursing care	0
Total TFC Score	**12 (Start and End of Study)**	

Table 3. N. Daniel Carragher Self- Reported TFC

Appendix O. HD Family Matters campaign

https://hdfamilymatters.com/