From Cradle to Grave: A Hermeneutic-Phenomenological Exploration of Living and Dying with Niemann-Pick Disease Type C.

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April 2019

Abstract

This thesis explores the lived experience across the lifespan of living and dying with Niemann-Pick disease type C (NPC). NPC is a neurodegenerative disease with no cure. One can become symptomatic at any age and the prognosis is often unclear. This emphasises the unpredictable nature of the disease and the wide-ranging impacts it can have on a person's life. Although work is being conducted into understanding the disease, there has been no research into understanding the illness experience. Incurable illness struggles to find a place in a society where the clinical gaze dominates and the acceptance of death is resisted. This thesis sought to understand the meanings associated with living with this rare disease at a lifeworld level. It is through such exploration that we can see the intimate links between self-body-world and understand such a critical existential issue.

Drawing on Heideggerian notions of at-homeness and homelessness, this series of studies focused on the existential nature of illness; the vulnerabilities and possible freedoms. Taking a multiperspectival approach to the interconnected lifeworlds of the person diagnosed and their family caregivers, three studies were conducted. The first explores living with NPC from a child's perspective; the second focuses on adults' experiences; and the last looks at end-of-life care and the dying experience for two families whose child has died from NPC. All three emphasised that illness has wider social and cultural implications that demand political and therapeutic intervention. From a re-analysis of these results, two quality of life scales for children and adults with NPC were developed and a reflective chapter on the novel use of phenomenology to develop items for these scales is presented. I make suggestions based on the results that a lifeworld understanding of these families' experiences is necessary within medical consultations, meaningfully engaging with what it means to be human.

Keywords: Rare disease, Illness experience, Phenonmenology, Lifeworld-led care, Well-being, Suffering

Dedicated to my Mum, Vera Aston, the most caring person I have ever known,
and in loving memory of my Dad, Robert Aston
27.08.1946-31.07.2010

Acknowledgements

Firstly I would like to thank both Actelion pharmaceutical company for funding this research and NPUK charity for the support that they have given to me throughout the whole research process. My sincere thanks go to all of the families who have bravely shared their stories with me. I feel privileged to have been able to listen to these experiences and this is something for which I will always be grateful.

Special thanks go to my two supervisors, Dr Rebecca Knibb and Dr Rachel Shaw. Thank you Rebecca for giving me this opportunity and for your support in helping me navigate the direction of where to take this programme of work. Rachel, thank you for the encouragement and wisdom that you have shared with me in moving this thesis forward in creative ways; this has been a really liberating experience and I am indebted to your patience and guidance.

Thank you to my family and friends who have spoken encouraging words to me, your support is something that I really cherish. A special thanks goes to Jenny Hammond who has accompanied me on this PhD journey. I am very thankful for our thousands of daily text exchanges and intense conversations on the meaning of life and death.

Huge thanks go to my Mum for her tireless efforts in keeping my offspring fed and watered in order for me to ‘get some work done’. I will always be grateful for your love and support for me during this challenging time.

Last but by no means least, my warmest thanks go to my husband, Jack, and our children, Molly and Jude. Jack, thank you for being such a constant source of support over the last few years. I am indebted to your kindness and patience. Molly and Jude, thank you for teaching me how to be efficient with time. I am very proud of you both. However, ‘screen time’ will be significantly less from now on - just so you know!

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Chapter 1 – Introduction

1.0 The story of this research

I will begin by telling the story of the genesis and development of this thesis. The PhD research was part of a larger project funded by Actelion, a pharmaceutical company, seeking to develop Quality of Life (QoL) scales for patients living in the United Kingdom (UK) with the rare, neurodegenerative condition, Niemann-Pick disease type C (NPC) (the disease trajectory is discussed in section 1.3). Actelion are the only pharmaceutical company to have an approved drug available for the treatment of neurological manifestations of NPC. There are currently no disease-specific QoL scales available for this condition. The project started with this task being its main outcome, yet as the PhD progressed and I grew to understand the illness experiences of people living with NPC, I began to question the assumptions and presuppositions of the meanings associated with QoL. In spite of the increasing popularity of QoL as an important component of outcome measures for those living with disease (Page et al., 2017) the application of a traditional QoL instrument to this study's population seemed inadequate to fully grasp the meaning of the lives of those with NPC. Furthermore, the value of a QoL instrument within a clinical setting seemed problematic due to the lack of available interventions that could be offered, considering the incurable nature of the condition. I began to search for a strategy that would transcend the traditional qualitative methods of scale development, i.e. thematic analysis or content analysis. To gain a deeper-rooted understanding of the human experience of living with a life limiting condition, and the 'biographic disruption' (Bury, 1982) that NPC can create, I realised we would first need to explore in-depth the first-hand subjective experiences of those diagnosed with NPC and their families. The intention therefore was to prioritise the concerns of people impacted by NPC by exploring the nature, and crucially the meanings they ascribe to their experience. Consequently, there was a huge amount of work to do *prior to* the development of QoL scales, which then came to dominate the programme of research reported in this thesis. It therefore became a pluralist endeavour involving phenomenological investigations of life with NPC involving, as one element of the work, the development of QoL scales.

Many instruments used to measure QoL imply that good QoL is the product of a lack or absence of difficulties rather than a favourable balance between positive and negative experiences (Cohen, Mount, Tomas, & Mount, 1996). For example, traditional indicators of poor QoL, such as limited mobility, may not actually preclude a sense of broader satisfaction or contentment in life. Life, and one's appreciation of it, is more complex than this, even and perhaps especially when one is ill. A disease may only provide a limited view of one's life experience. This alludes to the notion that the experience of being ill is a different phenomenon to the disease itself; indeed a distinction can be drawn between the illness experience and the disease. Carel (2016) discussed that: "Illness is the experience of disease, the 'what it is like' qualitative dimension as it is experienced and made meaningful by the ill person" (p.17). As Benner (1985) argued, the dual concerns with the experiential (health and illness) and the biological (disease) require different explanatory models; something the Western tradition has not necessarily consistently achieved. A preoccupation with a biomedical view of health has dominated medical discourse to the detriment of a fulsome understanding of the illness experience. When taking into account this disease versus illness discourse, it could be argued that QoL instruments focus more on the effects of disease as opposed to the illness experience, thus foregoing the acknowledgment of the myriad other complexities that people walk through when experiencing illness. A seemingly straightforward causal relationship is created without acknowledging any of the nuances that the illness experience has on life. Merleau-Ponty (1962) suggested that there is no adequate explanation of prediction, biological or philosophical, for the course of an illness. In other words, the disease trajectory, whether it be acute, chronic or terminal cannot predict the illness experience. As Carel (2016) articulated:

> *"Illness is a breathtakingly intense experience. It unsettles, and sometimes shatters, the most fundamental values and beliefs we hold. It is physically and emotionally draining. It can be physically and psychologically debilitating. Illness requires serious effort and continuous work to adapt practically to its limitations and to adjust psychologically to the pain, restricted horizons, and frustration it brings. It forces the ill person and those around her to confront mortality at its most direct and bare manifestation. In all of these ways illness requires labour, attention, and a conscious and sustained effort." (pp.3-4).*

It is important to understand the cause of disease and its physical manifestations for it to be placed in context. As discussed later, the heterogeneity of the onset of NPC symptoms and the nature of the symptoms themselves highlight the varied disease progression people experience. However, it is key to stress at this point that this study sought to understand the illness experience of living with NPC and not the 'facts' of disease. Exploring the illness experience from a first-person perspective therefore became the primary objective of this research, which required methodologies and theoretical frameworks that would allow one to understand experience in greater breadth and in greater depth than traditional QoL measures in order to fully understand the phenomenon of QoL when living with NPC. This approach would be founded on the meanings given to the illness experience and the rich complexities that arise from this, as opposed to describing the disease trajectory.

The intention of this introductory chapter is to outline and discuss the key components of this study and to provide a rationale for the impact this study offers substantively, theoretically and methodologically. The main focuses include:

1.1 The classification of a Life-Limiting Condition

1.2 Niemann-Pick disease type C: Background on what NPC is and its main effects

1.3 Quality of life: The concept of Quality of life and the problematic process of developing scales to measure this.

1.4 Phenomenology: The conceptual framework chosen to explore experiences of living with NPC.

1.5 Lifeworld-led care: The utilisation of this model to explore meaning and experience of living with illness.

1.1 Life-limiting condition

NPC is classed as a life-limiting condition (LLC). LLC is an umbrella term encompassing conditions for which there is no medical hope of cure, ultimately leading to death. Four types of LLCs have been identified (Hain, Devins, Hastings, & Noyes, 2013):

1. LLCs for which there is curative treatment that can fail (e.g. cancer, irreversible organ failures of the heart)
2. LLCs where premature death is inevitable (e.g. cystic fibrosis, muscular dystrophy)
3. Progressive LLCs without curative treatment options (e.g. NPC)
4. Non-progressive irreversible LLCs causing severe disability, health complications and premature death (e.g. cerebral palsy).

A LLC will shorten an individual's life, though they may continue to live active lives for many years (Hain, Heckford, & McCulloch, 2012; St Clare Hospice, n.d.). Some who are diagnosed with a LLC will spend a lot of time in hospital during their life and may have complex healthcare needs. However, the majority will live at home and be cared for on the whole by family members (Hunt, Coad, Staniszewska, Chesworth, & Chambers, 2015). This suggests that the care and support that people with LLCs might need, as well as their families and caregivers, is multifaceted and wide-ranging. As a consequence, co-ordination of care often falls to the parents or caregivers, which can be difficult to manage in terms of finding and being aware of help that is available (Hunt et al., 2015; Noyes. et al., 2013). The consequences this may have on the illness experience of people impacted by NPC have not been explored qualitatively before. Focusing on the illness experiences will provide a greater insight into the narrative of living with a LLC and the existential issues people may have due to the terminal nature of the diseases within this classification. This qualitative insight is a necessary prerequisite for a more detailed understanding of QoL throughout the life course with NPC.

1.2 Niemann-Pick Disease type C

NPC is a rare, genetic, neurodegenerative disease that can present during infancy, childhood or adulthood and is panethnic in its occurrence (Hendriksz et al., 2017). It is classed as a lysosomal lipid storage disorder, a metabolic disease that is characterised by a build-up of glycosphingolipids (fatty acids) in major organs, such as the brain, liver and other tissues (Hendriksz et al., 2017). The disease affects between 1:89,000 and 1:100,000 individuals (Mengel et al., 2013). However, its incidence is likely to be higher but the disease is under-recognised and misdiagnosed (Hendriksz et al., 2017). The main effects of the disease are neurological and visceral symptoms (Patterson et al., 2013). The visceral symptoms include prolonged neonatal jaundice and cholestatic symptoms, such as nausea, pain in the abdomen and inability to digest certain foods (Hendriksz et al., 2017). Psychiatric symptoms are often present including behavioural problems, psychosis, bipolar disorder and developmental delay (Hendriksz et al., 2017). Often, patients are diagnosed in early years with one or more neurological manifestations such as supranuclear vertical gaze palsy (i.e. difficulty with upward and downward eye movement), cerebellar ataxia (i.e. unsteadiness of gait in turn causing problems with walking and clumsiness), dysarthria (i.e. problems with speech) dysphagia (i.e. problems with swallowing) and learning problems (Patterson et al., 2013). The clinical features of NPC include progressive loss of vision, hearing, muscle co-ordination and mobility. Prognosis varies but patients who display neurological symptoms in childhood have been shown to deteriorate faster compared to those who become symptomatic later on in life (Vanier, 2014). In addition, cognitive decline leading to early onset frontotemporal dementia can occur resulting in premature death in most people diagnosed (Patterson et al., 2013). However, the clinical profiles of people with NPC are heterogeneous as well as disease onset and progression.

Due to the rarity of NPC, the diagnosis process can be prolonged, complex and stressful with diagnoses usually being given 5-10 years after initial symptom onset (Burlina, 2014; Mengel et al., 2013; Patterson et al., 2012, 2013). One of the reasons for this is the unrecognised nature of NPC due its rarity and the lack of awareness clinicians have of the disease. A number of referrals are made within primary care and across a range of medical specialities due to the unrecognised symptoms of NPC among clinicians. Specialities such as paediatrics, adult neurology, haematology and psychiatry are some to which referrals are made, thus demonstrating the heterogeneity of symptoms and specialist needs that are required (Hendriksz et al., 2017). This in turn can lengthen the diagnosis period and subsequent intervention. There has been an improvement in the availability of clinical screening tools (the NPC suspicion index) and genetic analysis methods that improve the detection of NPC (Hendriksz et al., 2017). Yet, the broad spectrum of the manifestations and the variable age of onset highlights the complex nature of the disease and the lengthy diagnostic process patients and their families endure (Burlina, 2014).

Currently, there is no cure for NPC and the only approved disease-specific drug available is Miglustat (Zavesca®; Actelion Pharmaceuticals Ltd), which was approved in 2009 (Pineda, Walterfang, & Patterson, 2018). Miglustat is offered to children and adults with NPC for the treatment of progressive neurological manifestations (Patterson et al., 2013). Some studies indicate that Miglustat slows the progression or delays the onset of neurological symptoms (Di Rocco, Dardis, Madeo, Barone, & Fiumara, 2012; Patterson et al., 2015; Skorpen, Helland, & Tennøe, 2012). However, most studies are prospective rather than randomised controlled trials, due to the challenges both in terms of design and finance in execution (Pineda et al., 2018). Not all patients with NPC will benefit from taking Miglustat and some refuse to take it due to the side effects, the most common being diarrhoea (Lyseng-Williamson, 2014).

Overall, the disease trajectory of NPC is varied in terms of onset of symptoms, severity of symptoms and disease progression. This suggests that people's experiences of living and ultimately dying with this disease will be mixed and complex, further justifying the need for an idiographic approach to understand the experience of living with NPC in more detail.

1.3 Quality of Life, instruments and scale development

The concept of QoL is a highly valued focus of research and consideration within clinical practice. Despite this popularity, the lack of consensus on the definition of QoL is a source of confusion. Kassianos (2015) commented that "QoL is among the most inconsistent concepts in the literature, and very often a reader needs to see and decide how the author(s) define QoL." (p. 1193). Surprisingly, this is often difficult to ascertain within research papers as a definition of the term is not provided, which assumes a consensus which does not exist. Arguably, one of the most respected and recognised definitions of QoL is the World Health Organisation's (WHO) which describes it as being: "individuals' perceptions of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards and concerns" ("The World Health Organization quality of life assessment (WHOQOL): Position paper from the World Health Organization," 1995: p.1404). It is a therefore a multi-dimensional concept and is understood to be "affected in a complex way by the person's physical health, psychological state, level of independence, social relationships, personal beliefs and their relationship to salient features of their environment" (World Health Organization, 1998: p.1). From the literature concerning measurements of QoL, the key domains that tend to be assessed are: physical, mental or social health; social interactions; economic/vocational status and religious or spiritual issues, thus complementing the WHO's definition of QoL (Kassianos, 2015).

Health Related Quality of Life (HRQoL) is similarly inconsistent both in its definition and its operationalisation (Eiser & Morse, 2001; Waters, Maher, Salmon, Reddihough, & Boyd, 2005). A more simplistic definition conceptualised by Spilker and Revicki (1996) sees HRQoL as a subset of QoL, referring to an individual's perception of their health, which according to the WHO, is made up of physical, mental and social well-being. Many references to both QoL and HRQoL are found in health care and patient-reported outcome measures. These serve to enable Healthcare Professionals (HCPs) to better engage with patient wellbeing, providing a useful and efficient method of measuring the impact of disease or health status on an individual's QoL (Davis et al., 2010). These can then be used to tailor interventions for a patient or a carer or more widely in informing health policies. Within clinical research, there is a recognition that QoL should be taken into consideration and it is therefore in the position of receiving substantial amounts of money to fund projects that develop QoL scales for specific diseases, for example see the C-DEMQOL (Alzheimer's Society, n.d.; Fitzpatrick, Fletcher, & Gore, 1992). However, the aforementioned confusion over definition and practice with this concept is potentially leading to a culture whereby lip-service is granted to the notion of QoL without proper engagement with individual experiences and the nuances they convey. Cohen et al. (1996) argued that the inclusion of QoL measurement in clinical trials research is a reaction to acknowledging the need to implement a wider professional goal: "the alleviation of suffering, rather than the narrower goal of fighting disease" (p. 576). The degree to which these measures fulfil their function is open to debate, yet one must recognise that there is an increased emphasis on the broad concept of QoL and its enlarged influence on treatment and policy despite its vague definition:

> *"We find ethical philosophers suggesting that decisions about the termination of human life ought to be based on quality of life criteria (Kuhse, 1987), physicians who claim that they can measure the impact of a particular therapy on the quality of life of their patients (Croog et al., 1986; Evans et al., 1985; Klonoff et al., 1986), and health economists who think that quality of life considerations should influence the allocation of financial resources within the health service (Williams 1986)." (Draper, 1992: p.965).*

Separate to the more specialised definitions and uses within the clinical and research worlds, the phrase QoL is used within everyday conversation. This is positive in the sense of a general recognition of the concept for the wider population, but there exists a further dilution of meaning through its frequent usage, contributing to the epistemological inconsistency observed in the literature and beyond. The roots of attempts to define QoL can be found in the Aristotelian notion of *Eudaimonia*, meaning happiness or fulfilment as a feeling and activity (Fayers & Machin, 2007). It is of course a subjective concept that individuals will imbue with their own meaning as is recognised in the WHO's definition: "individual's perceptions…". In tune with this, Rotstein, Barak, Noy and Achiron (2000) argued that there are two ways of understanding the assessment of QoL; the objective and the subjective. The objective relates to people's universal requirements for living including their access to food, shelter, and good health. Whereas the subjective is concerned with the inner experiences of the individual, which Rotstein et al. (2000) dichotomised as positive and negative dimensions. Calman (1984) suggested that QoL must include all areas of life and experience and take into account the impact of illness and treatment.

In relation to the pragmatic use of understanding QoL within the context of illness, QoL scales are seen to serve as a way of capturing the broader impacts of living with disease. Their most practical use is in clinical trials to measure the effect of an intervention (Fitzpatrick et al., 1992). However, the way in which these tools are developed and the evidence that is used to suggest that these are the broad impacts to be measuring is uncertain (Lim & Zebrack, 2004). As discussed earlier in the chapter, it is possible for these QoL measurements, especially HRQoL tools, to be reductionist in their focus on the disease trajectory. The physical domain in QoL instruments is usually overemphasised and other domains underrepresented (Carlozzi et al., 2016; Cohen et al., 1996). When considering the use of QoL measurements it is important that these tools do not detract from the illness experience in its wholeness or merely focus on this as a secondary outcome of disease. The seemingly fragmented components that are traditionally seen to comprise of what makes QoL, and subsequently what are used in QoL measurements, such as happiness or physical ability, may fail to acknowledge the totality of human existence. An approach that is more considerate of the illness biography of an individual rather than on the disease progression and its physical manifestations may align itself more with what 'quality' of life means from an ontological perspective. With this said, it is important to take a step back and consider the way in which these QoL scales are developed in terms of the evidence used and the methodological approaches taken to generate items that claim to understand and measure QoL when living with a specific disease.

There are two types of instruments to measure QoL that are commonly used: generic and disease-specific. Generic scales are intended to be applicable to all population subgroups and useful for comparing outcomes of subgroups (Bjornson & McLaughlin, 2003). Disease-specific scales have been developed to be applicable to one group and are useful to detect changes in a condition (Bjornson & McLaughlin, 2003; Fitzpatrick et al., 1992). A review of the literature on disease-specific QoL scale development, specifically within the context of neurodegenerative diseases, found that a common methodology is to use in-depth interviews with the population group followed by a thematic analysis (Parkinson's Disease, PDQ-Carer; Jenkinson et al., 2012) or grounded theory (Cerebral Palsy, CPQOL; Waters et al., 2005) to develop the scale items. One study did not provide the type of analysis they used to develop items, although techniques from both thematic and content analysis are implied (Parkinsonism, PQoL; Pillas et al., 2016). Some items have been developed based only on clinical impression from observation work or focus groups with patients and caregivers and literature reviews (Multiple Sclerosis, CAREQOL-MS: Benito-León et al., 2011; Cohen et al., 1996). One study used a combination of the methods listed above (Huntington's Disease, HDQOL-C; Aubeeluck & Buchanan, 2007). Very little detail is provided as to the development of these scales, with reports prioritising the validation over the developmental process. Although, overall good internal consistency has been found in the above instruments (Page et al., 2017), key factors of the illness experience are missing. The weak reporting of methodological approaches used in the development process make it difficult to discern what level of detail was generated in the qualitative data and the analysis.

Arguably, a stronger methodology for the developmental process would provide a more robust basis for QoL scales that is based on lived experience. Further strengthening to this approach would be to draw on theoretical conceptualisations of human phenomena that aim to provide the fullest explanation possible as to what it is to live with illness. Benner (1985) argued that "QoL can be approached from the perspective of being, and does not need to be approached merely from the perspective of doing and achieving" (p.5). Understanding the 'quality' of life should be within the context of understanding what it is to be human. It could be argued that the methods used to develop QoL scales are not getting to the crux of what they are intended to measure due to the lack of attention being given to meaning-making and exploration of key phenomena. As Draper (1992) wrote: "The life whose quality is in question is not some theoretical principle, but always and in every case the characteristic of human being" (p.966). There is a greater depth to what constitutes life in all of its quality from an ontological perspective, an issue deeply embedded in what it means to be human.

With this in mind, the nature of exploring QoL for people living with NPC necessitates an engagement with broader philosophical constructs and frameworks that can be used to understand this phenomenon. Much of the need for such engagement with theory is driven by the desire to maximise the conception of what constitutes QoL and the need to push beyond a narrow clinical definition. An interaction with other theories is required that seeks to expand the scope of enquiry and ask more fundamental questions about how human beings understand the world about them. There is a need to broaden the lens when considering QoL, to step back and consider whether such measures, traditionally conceived, give us a real impression of an individual's understanding of their life and experiences. Of course, close examination of experiences and the degree to which these can be translated into a tool that can be used to measure 'quality' of life runs the risk of measuring specific factors that contribute to a very individualised view of QoL rather than assessing a more general reflection. An attempt to generate fully a comprehensive list of factors that affect QoL would not be possible (Cohen et al., 1996). However, with regards to this study, the rarity of NPC may allow for an experiential approach to item development which may resonate closely with a large proportion of the population. Underpinning this developmental process with philosophical insight sought to give a more meaningful foundation of 'what it is to be a human living with NPC'. This is over and above what other scales have offered thus far within the context of neurodegenerative conditions. The next section will discuss phenomenology as a philosophical framework for this programme of research.

1.4 Phenomenology as an approach to inform study

Phenomenology is a movement within philosophy that studies human experience (Taylor, 1993). Phenomenology focuses on the examination and description of perceptions of meanings and experiences for the individual; the interface between consciousness and the real world (Husserl, 1954/1970; Moran, 2000). In general, phenomenology centres around "embodied, experiential meanings aiming for a fresh, complex, rich description of a phenomenon as it is concretely lived." (Finlay, 2009: p.6). Langdridge (2007) defined phenomenology as a discipline that has as its aim the study of people's perceptions and the meanings attached to them. Phenomenology as a theoretical framework has been taken on by psychology as a conceptual approach for research to understand the human project. The origins of phenomenology are found in the work of Husserl (1859-1938) and Heidegger (1889-1976), among others. It finds expression in the ontological, individualist framing that seeks to understand the essence of human experience. This essence, which is sought by phenomenological approaches, is understood as the individual's meanings found in shared experience (Patton & Patton, 2002) and it is the collation on the study of essences (Van Manen, 1990) that drives phenomenology towards an understanding of the human experience.

As outlined above, this programme of research was designed to develop an understanding of the meaning of living and dying with NPC and centres its focus on the existential line of phenomenology, which is concerned with pre-reflective everyday life and human experience, developed from the philosophical work of Heidegger and Merleau-Ponty (1908-1961). Phenomenology is apt for the philosophical study of illness in that it is a useful methodology for describing and ordering experience, therefore it can be applied to the first-person view of illness, in order to illuminate this experience. This approach allowed for a deeper understanding of the existential meanings of living with NPC and the potential features of a QoL instrument that would seek to understand the meanings those people attribute to their experiences. This is especially valuable when working with such a rare condition because it then means that these meaningful experiences, rather than preconceived ideas and concepts from other QoL scales, are used as the starting point for an instrument that can then be used in clinical and research practice. The rooting of the scale development in experiences of life with NPC gives epistemological validity to the process and moves ever closer to the humanisation of clinical practice (Galvin et al., 2018).

1.5 Lifeworld-led care

Galvin and Todres' work on Lifeworld-led care provided a rich conceptual backdrop to this work because it marries the phenomenological theory to the clinical application. This approach is based on the phenomenological notion of the lifeworld (*lebenswelt).* Husserl (1936) suggested that in order to understand human experience, we must explore the lifeworld. Building on the tradition of Svenaeus (2000b) and Toombs (1992) Galvin and Todres' aim has been to use philosophical frameworks to provide a firmer basis for understanding health and care issues. Todres, Galvin and Dahlberg (2007) describe the lifeworld as the following:

> *"We as humans may find it difficult to notice and articulate the humanly qualitative nature of the world we live in. Ours is not a universe of neutral objects with ourselves fitting in to these physical forces; nor are we simply minds or brains that attach meaning to this neutral world of processes, others and things. Instead, there may be something more humanly intimate about the world in which we find ourselves. The nature of human-world intimacy was named the lifeworld, the beginning place-flow from which we divide up our experiences into more abstract categories and names… The primary nature of this relational reality means that there is no objective world in itself, nor an inner, subjective world in itself; there is only a world-to-consciousness." (Todres, Galvin, & Dahlberg, 2007: p.55).*

To help generate an understanding of the Lifeworld a number of dimensions have been identified that allow for a fuller description of human experience: temporality, spatiality, intersubjectivity, embodiment, identity and mood. This complex web of interrelated constituent parts has been variously explored and analysed by Husserl (1936 [1970]), Heidegger (1927 [1962]) and Merleau-Ponty (1945 [1962]) and whilst there are differences in their work, each includes the aforementioned dimensions in some way. Todres et al.'s (2007) Lifeworld-led approach to care enables us to take the components both separately and as a whole to create a framework for understanding the lifeworld of individuals and, in turn, how this could be used to drive a fuller understanding of care. These will be explored in chapter 3.

The lifeworld is understood as an experienced world of meaning. As Todres et al. (2007) commented: "Any qualitative moment or event is part of a larger story and place; every word is said in relation to other words and meanings" (pp.55-56). This delicate web of meaning and experience on an individual level has not been fully exploited in the context of scale development. Understanding more of the fullness of living with NPC within these dimensions may have the broader advantage of advancing healthcare and policies that better engage with the illness experience in a way that humanises care (Galvin et al., 2018). In relation to this lifeworld-led approach, this study therefore sought to contribute breadth – and depth - to the evidence base of living with illness at a lifeworld level. People's expressions of their experiences of health and illness, their shared and individual journeys and their interactions with each other presented a new meaning of what it is to be human and live with NPC.

1.6 Research aims and objectives

No qualitative work in the UK has been conducted looking at the experiences of living with NPC, which raises questions regarding the level of support and nature of interventions people with this disease receive. Generic QoL scales are being used to measure the effects of drug trials on people living with NPC. This means that the more specific complexities that may come from living with a rare disease are not being taken into consideration. Prior to developing a disease-specific scale to measure QoL, explanations in the phenomenal realms of health and illness must be explored before adequate descriptions and predictions of living with disease can be developed (Benner, 1985). With the conceptual framework of phenomenology in conjunction with the lifeworld theory, this programme of work will interpret 'quality' as being that which helps or hinders people from living an authentic life. Any disruptions to a person's lifeworld will be seen as a key avenue of exploration in how living with NPC may impact upon a person's QoL. Understanding the lived experiences of someone with NPC and the impact the illness experience has on their QoL may help to advance interventions aimed at modifying factors that are seen to disrupt the lifeworld, thus being sensitive to what really matters to people living with NPC. In addition, exploring end-of-life care and the dying experience of someone with NPC will contribute to the entirety of the illness journey and illuminate the impact to the lifeworld that exists when faced with death. Doing this will contribute to the empirical evidence base for the provision of palliative care within rare disease. Consequently, the overarching aim of the work presented in this research programme was to explore the meaning of living with NPC across the lifespan of the illness experience. The objectives are to:

1. Explore the lifespan of living with NPC from first-person experience: children, adults and dying with NPC.
2. Deepen understanding of the illness experience from diagnosis onwards and how it impacts upon the life of the person with NPC and their family members.
3. To consider how data generated from this study could be translated into a tool that measures QoL. The use and value of this instrument in clinical service provision will be considered.

1.7 Outline of thesis

The chapters of this thesis seek to tell the story of living with NPC across the lifespan. This will be addressed within the following chapters.

Chapter 2 provides an overview of the holistic experience of living with a LLC within the context of existing research on living with rare disease. The first part of the review will focus on synthesising research that has explored the experience of living with rare diseases on the person and the family carers. The second part of the review will focus on the current policies that are in place for care provision of people living with a LLC in the UK. This section of the review will also look at research that explores HCPs' experiences of caring for people impacted by a LLC, with attention being given to communication and decision-making, especially in the context of end-of-life care surrounding withholding and withdrawing treatment.

Chapter 3 outlines the methodological approaches taken in the three empirical studies included in this thesis. A rationale will be provided for the chosen logic of inquiry that frames these studies within the phenomenological worldview I chose for this research. Further details on the methods chosen and the analytical approaches taken are provided.

Chapter 4 is the first of the empirical studies. This chapter utilises a multiperspectival approach and explores the experiences of children living with NPC through the lens of their parents' meaning-making. It seeks to illuminate the disruptions that the illness experience has on the lifeworld and its intimate links to body-self-world.

Chapter 5 moves on to explore the experiences of adults living with NPC. All participants were diagnosed during adulthood and so the comparison between the life they once knew and the life they now lived with illness is illuminated. The notion of narrative wreckage as antecedents to a potential sense of homecoming are important within this chapter.

Chapter 6 finishes the exploration of the lifespan by looking at the dying experience of two families whose child died from NPC. This chapter looks at end-of-life care, death and life afterwards for the parents. As part of the results, an embodied interpretation is presented to evocatively capture the experiences of both families, providing an empathic, aesthetic phenomenological reflection of the death of a child.

Chapter 7 presents the development of two quality of life scales for NPC. This is a reflective chapter, which discusses the novel approach taken to develop the scales using phenomenology. It reflects on the use of QoL scales for people living with NPC and whether these tools provide an appropriate way of understanding the holistic illness experience.

Chapter 8 concludes the thesis by discussing the key findings from the empirical studies and the implications that these have on clinical practice and research. A discussion of the strengths and limitations as well as a reflective discussion of my personal account of conducting this research will be provided.

Chapter 2 – The holistic experience of living with a life-limiting condition: a review of the literature

2.0 Introduction

Following on from the introductory chapter outlining the key areas of focus for this doctoral research, it is important to understand the existing literature to which this PhD is contributing. This chapter will be divided into two parts. The first part will outline research that has looked at the illness experiences of living with a LLC both from the personal perspective of the one diagnosed and the family unit. Studies that look at how illness impacts more widely on relationships in society will be explored as well as a more specific focus on support within healthcare services, which will be used to understand in more detail the illness experience. The second part of the review will explore the care provision available for people living with a LLC in the UK, critically assessing guidelines that are in place for HCPs to adhere to when caring for someone with a LLC. A review of the evidence surrounding HCPs' experiences caring for those with a LLC will be discussed, focusing on communication and decision-making. This will include the ethical dilemmas of care surrounding withholding and withdrawing treatment.

2.1 Search strategy

A search of the available literature was conducted in the following databases: Web of Science, Science Direct, Opid Medline, and Google Scholar. The initial search terms used were 'life-limiting illness' and 'life-limiting condition'. These were used in order to generate a broad range of studies that included multiple perspectives of living with or caring for someone diagnosed with a LLC. Abstracts from articles identified were screened for relevance and reference chaining of included studies was conducted. Additional searches were conducted to capture the current state of affairs regarding policies for healthcare services in caring for people with a LLC. Thus this review included publications, commentaries and reports from some international guidance but predominantly from national health governing bodies. In addition to this, editorials and perspective pieces from HCPs and researchers specialising in palliative care were included.

2.2 Introducing a review of the illness experience

Being diagnosed and living with a LLC causes significant ‘disruption’ to the individual’s life as well as to the family unit (Bury, 1982; Hurt, Cleanthous, & Newman, 2017). Bury's (1982) concept of biographical disruption suggests that an individual’s sense of self and daily life are disrupted by the illness experience (Felde, 2011). This notion is given brutal expression in Frank’s (1995) conception of illness as a ‘narrative wreckage’ which imposes an irrevocably altered sense of being. This narrative wreckage can be seen in Carel's (2013) description of receiving her diagnosis with a LLC:

> *“Pain and fear struck like a physical blow. It is difficult to describe the physicality of bad news. I remember looking at the room and feeling confused: it looked the same, while my life had been turned upside down. Make it stop, I thought. This is the wrong story. Someone come and fix it. Someone do something. The realisation that everything was about to change, that a new era was about to begin, seared like burning oil on skin.” (p.4).*

The illness experience that Carel (2013) describes, beginning at diagnosis, is life-altering and causes people to reflect on the sense of loss. Loss is not only found in the objective bodily deterioration and complex care needs that can come as a result of disease progression but is also experienced in the loss of opportunities and possibilities, forcing one to rethink the goals and expectations once held (Carel, 2017). This is especially magnified in LLCs due to their incurable nature and uncertain prognosis.

Quite often in the existing literature, the illness experiences of the person diagnosed is intertwined with those of their family members. This is congruent with the notion that the lifeworlds of those most involved are intimately shared, and are thus disrupted and impacted upon relative to one another. The following subsection will initially prioritise research that has looked at the individual who has been diagnosed with a LLC in order to draw on the distinct voice of the illness experience. It will then move on to review research that has looked at families’ experiences of caring for someone with a LLC in order to separate the perspectives and understand more fully the nuances of these alternative positions. The reviewed literature will be presented thematically and as it develops, the scope will broaden to include an appreciation of the individual, familial, healthcare and societal implications and perspectives that must be brought to bear in order to comprehend the interrelated and interconnected nature of the social world in which we live.

2.2.1 The illness experience of the individual

Throughout the illness trajectory, existential thoughts and concerns surrounding life and death can be present (Harris, 2015). Sloan, BrintzenhofeSzoc, Mistretta, Cheng and Berger (2017) explored the meaning-making of individuals who had been diagnosed with a chronic or LLC illness. Participants were aged between 26-65 years and took part in interviews. Themes that emerged from the data indicated that significance and purpose were found after receiving their diagnosis especially in terms of the meaning they placed on relationships with friends and family. They also found that their illness experience had given them a greater appreciation of the life yet to be lived.

An issue that was not addressed sufficiently by Sloan et al. (2017) was the definitive difference between living with an incurable illness and a chronic illness. The distinction between the two 'categories' of illness may suggest a key variation in meaning-making. The incurable nature of the LLC may impact greatly on one's temporal experience in living with the knowledge that the disease cannot be cured. On the contrary, living with a chronic disease may not bring this immediate level of temporal concern of death but may instead be difficult in terms of reckoning with the continuous and long-term nature of the disease. The heterogeneity of including participants within two different categories of conditions does raise concern regarding the meaning of the results especially when this had not been acknowledged by the authors. The merging and generalisation of distinct illness pathways limits the overall conclusions of this study.

Cook, Jack, Siden, Thabane, and Browne (2016) investigated the aspirations of young adults who were living with LLCs by interviewing ten young adults. The participants in this study were aged between 19 and 29 years where a prognosis of surviving until their early 30s was given. Although the overarching theme found from the interview data was that of living with uncertainty, for some this uncertainty encouraged them to focus on the things in life that they perceived to be achievable and desirable. These aspirations mainly included postsecondary education and vocational courses, through which some conceived of future success. Some of the young adults worked in relatively short-term timeframes, a mindset developed due to the insecurity of their future. This led to an outlook characterised by a preoccupation with the immediate and the present, whereby young adults had recognised the degenerative nature of their illness and thus sought to live in the moment. This study appears to be among the most recent to explore the goals of young people living with a LLC, with its particular emphasis on the meanings they attributed to their experiences of both the present and the future. The study included people living with a variety of LLCs, without making reference to the idiosyncratic accounts of each illness experience in the results. As well as this, the varying temporal dimension of the experiences was not taken into consideration, such as those who were expected to live for another decade compared to those who might only live for a further two years. These distinct differences are important to consider especially in the context of the ‘aspirations’ that are being explored.

In addition to this, Cook et al. (2016) commented that some young adults could not take part in the study due to illness decline. It would have been beneficial to seek their parents’ and/or caregivers’ interpretation of the aspirations of the young people in question. A potential means of incorporating the voices of those whose illness precludes their traditional means of participation would be to adopt an interpretative phenomenological approach to data collection and analysis. This would provide an opportunity to interpret how young adults may experience and understand their illness, through their families’ interpretations of this. Adopting phenomenologically-based analytical techniques means that we can attempt to understand the experiences of people living with illness through the people who know them the best. If a young person is unable to communicate due to illness, a relational understanding of how they make sense of their experience through the families’ meaningful interpretation of their mood, facial expressions and body language could enable an important voice to be heard (Smith & Shaw, 2016).

Johnston, Jindal-Snape and Pringle (2016a) conducted a systematic review to identify articles that aimed to understand the way that young adults with a LLC experienced life transitions. Studies that looked at adolescents and young people aged 10-25 years with a LLC were included. Eighteen studies were included in the review and findings were thematised into two key transitions for young adults, which were the ‘illness transitions’ and ‘developmental transitions’. In the ‘illness transition’ the illness trajectory and coping strategies were discussed. The impact that ‘illness transitions’ had comprised of adaptations that needed to be made in terms of lifestyle changes, including house renovations and changes to employment, reflecting the more practical implications of living with a LLC. Feelings of grief and the loss of the life they once knew were found in 8 out of 18 studies. Social isolation and strains on relationships were found, illuminating the disruptions caused to the intersubjective experiences of individuals.

‘Developmental transitions’ for young adults included the impact of illness on personal development, the likelihood of employment and engagement in further education. Studies showed that young adults anticipated obstacles because of their condition. Continuing and/or developing new relationships was shown to be challenging as participants of all ages expressed difficulties with their social life due to their illness. This was especially apparent when their peers were going to university. The concern of burdening future relationships due to the young person's illness was also something that caused apprehension. Thus the stunted nature of established relationships and the uncertain perception of the nature of future relationships was a key finding. It is unclear as to which methodologies were included in Johnston et al.'s (2016a) review. In addition, how they synthesised the data from the studies they included is uncertain, thus calling into question the transparency of this review.

Johnston, Jindal-Snape, Pringle, et al. (2016b) also explored experiences of young adults living with a LLC as they moved into adulthood. Participants were aged between 17 and 23 years and were recruited through their admittance to children's hospice services. Young adults felt isolated and alienated as peers were pursuing avenues that the young adults could not. This led to some feeling like they had no life experience and were "wrapped in cotton wool" (p.8). The ambiguous illness trajectory meant that young adults lived with loss, a loss of what they anticipated the future bringing, as well as the associated loss of physical enablement. Similar to Cook et al. (2016) some participants could not take part in this study due to ill health. By not including a multiperspectival (Larkin, Shaw, & Flowers, 2018) approach these participants' experiences are consequently not conveyed. Again, including multiple LLCs presumes homogeneity but for each illness the experiences will be different and this needs to be acknowledged within the results.

The impact of living with and dying from a LLC can be strongly associated with emotional distress for the person diagnosed and for the family (Penman & Ellis, 2015). Penman and Ellis (2015) examined the range of emotions experienced by people living with a LLC. They found that the emotional response that was the most challenging when living with a LLC was fear, especially when dying. The use of pain relief in dying is a key priority which HCPs adhere to (NICE, 2017b; Mannix, 2017). This aids relief from an objective point of view, but more existential fears such as what happens after death, how it feels when one is dying, and how loved ones will manage when death occurs are very difficult to manage and allay (Penman & Ellis, 2015).

Penman and Ellis (2015) found that fears expressed by people who were dying consisted of: falling, turning into nothingness, not being comfortable, dying and dying alone, leaving children behind. Strategies to address fears were also explored such as communicating about their fears, involving HCPs and more spiritual approaches such as praying and talking to religious leaders. Penman and Ellis (2015) argued that these emotions have a purpose to help reach an adaptation to assist in managing and coping with the turbulent change in realities. Unlike other studies, the focus was placed on the emotions felt by people who were in the final stages of life, which gives important insight into the individual's perception of dying. Arguably this approach needs to be extended across the lived experience of the life cycle in order to understand the nuances of life with a LLC, including the origins and evolution of the emotions felt most prevalently when facing death. Garnering an idiographic, in-depth understanding of the illness experience is the first step in developing suitable interventions, which support people at the end of their life.

Overall, research focusing on the meanings ascribed by the individual to the illness experience revealed a sense of uncertainty as to what the future will bring, the significance of fear as a key emotion and the personal growth that can come with living with a LLC. The elements of temporality and relationality meet with existential issues attached to concerns surrounding identity, death and dying. There is a paucity of rich descriptions of the illness experience from people diagnosed with a LLC in the literature, especially from the perspective of children. There is a need to concentrate on understanding the nuanced, everyday lived experiences of living with a LLC with epistemologically-led methods that are more centred on the idiographic nature of exploring the meanings associated with living with incurable illness. This review will now focus more on the impact that living with a LLC has on close family members and the illness experiences that are shared within the family unit.

2.2.2 The families' experience of living with a LLC

Living with illness is not simply a contained experience for the person diagnosed but also impacts closely upon family members (Lane & Mason, 2014). Kars, Grypdonck and Delden (2011) explored the experiences of parents who had a child with advanced, incurable cancer and described the diagnosis as being the "collapse of their parenthood" (p.262). Ware & Raval (2007) interviewed fathers of children who had a LLC, the diagnosis of which left them feeling "powerless" (p.554), with their world now feeling unsafe and insecure. Jordan, Price, and Prior (2015) similarly described living in the knowledge of the finality of life and certainty of death as a land of limbo for their child; an isolated existence between life, as it is conceived of by others, and death.

When diagnosed with a LLC, research has shown that the family system alters in order to accommodate the complex care needs of the person diagnosed (Nicholl, 2012; Nuutila & Salanterä, 2006; Williams, Ayres, Specht, Sparbel, & Klimek, 2009). The implications of this have been found to affect employment, where a parent may need to give up work. This then brings with it the associated financial repercussions and stretching of resources (Runswick-Cole, 2010). Due to the progressive nature of LLCs, the deterioration of people diagnosed will mean an increasing dependence on parents/spouse/carers (Brennan, Hugh-Jones, & Aldridge, 2013).

The role of parents has been found to change when caring for a child with a LLC (Persson & Sundin, 2008). The day-to-day care ranging from the physical needs of the child to any behavioural or psychological ramifications of the illness have been found to be challenging for parents to manage (Nicholl, 2012; Nuutila & Salanterä, 2006). Whiting (2014) explored parental perceptions in caring for a child with a LLC. One of the mothers interviewed reflected that "*you stop being just a mum*" (p.29). This impacts upon the parents' sense of identity *as parents*, now having different roles and responsibilities to manage (Whiting, 2014). For example, being a parent, a partner, a carer and a healthcare 'professional' are jobs that come with the complex care needs of the individual (Kars et al., 2011). This might include giving medication and attending to personal care, roles for which feelings of being ill-equipped to do so were expressed (Kars, Duijnstee, Pool, Van Delden, & Grypdonck, 2008). In spite of this, research has also suggested that there is a degree of nuance in the changing relationship between carer and cared-for. Persson and Sundin (2008) commented:

> *"The onset of this new role may be difficult for the caregiver to determine because there is no clear distinction between the experience from an ordinary caring relationship within the family and that from family caregiving" (p.380).*

For parents of children with a LLC, the irrevocable nature of the illness trajectory and the longevity attached to the care needs has been shown to impact upon the psychological health of those involved. Parents' mood has been found to fluctuate due to feelings of frustration over the relentless nature of care; this is accompanied by periods of anxiety and depression as well as feelings of fatigue and sleep disruption due to the physical impact of caregiving (Lane & Mason, 2014; Steele & Davies, 2006). Steele and Davies (2006) interviewed parents of children with a LLC and found some parents were clinically depressed, with some experiencing suicidal thoughts or attempting suicide.

Persson and Sundin (2008) interviewed the significant others (i.e. relatives or friends) of a person diagnosed with inoperable lung cancer, in order to gather the meanings attached to the significant others' lived experiences. Data were analysed using a hermeneutic phenomenological approach. The unexpectedness of caring for their significant other led to them feeling dislocated from the world they knew, a world now replaced by the darkness of forthcoming loss. As well as this, the relationships no longer felt symbiotic or equitable as the person started to drift away from being *able*. This dislocation was fought against by the significant other in making the effort to manage the illness well and 'endure the situation'. This incorporated trying to live in the present whilst living in the shadow of future loss. This study provided an in-depth, rich understanding of the existential challenges that caregivers face that have not been dealt with as fulsomely in other studies. Focusing on the lived experiences of caring provided a rich understanding of the complexities of illness by exploring the subjective meanings.

Jordan et al. (2015) interviewed parents who had cared for their child with a LLC until they had died, in order to understand their experiences of caregiving throughout the illness trajectory. Thematic content analysis was employed. Parents commented that when they were caring for their child they felt suspended in a world separate from the one they lived in prior to their child's diagnosis and separate from the one that they will live in when their child dies. For some this was a conscious choice, to not live in the 'world of the well' as they no longer felt a sense of belonging there. The homogeneity of the sample meant that a variety of experiences were analysed together. For example, 5 out of the 15 parents interviewed had children who had died before the child had reached the age of 1 year. Only one parent's child had died between the ages of 1-10 years and 9 between the ages of 10-18 years. Considering this study was exploring the experiences across the entire illness trajectory, no other information was provided in terms of the age at which a diagnosis was given and whether the disease progression had occurred very quickly or over the course of years. This raises questions as to whether all of the experiences across the sample should have been analysed together due to the varied nature of experiences across age groups. Refining the focus of the study to one age bracket may have provided a closer, more in-depth understanding of the families' experiences.

Research has found younger family members take on greater responsibility than would be expected ordinarily (Brennan et al., 2013). Read, Kinali, Muntoni and Garralda (2010) documented psychosocial adjustment in teenage siblings of children diagnosed with the LLC Duchenne muscular dystrophy (DMD). Associations between the emotional and psychological adjustments of unaffected siblings during the illness progression were observed. This was especially so if there was a closeness in age between siblings and the person diagnosed with the LLC. Read et al. (2010) found that unaffected siblings experienced an increased risk of emotional problems but did not provide information regarding the nature of these emotional problems. This lack of description was related to the methods used in the study, which required participants to complete a battery of questionnaires measuring well-being, general health, anxiety, depression and family care giver burden. These do provide a general scope as to how participants with an ill sibling *feel*, but do not give insight into the range and depth of emotion experienced, nor the contextual awareness that can enlighten understanding further.

Brennan et al. (2013) used grounded theory to explore the experiences of siblings of children who had a LLC and the ways in which they coped. The three themes generated were unified under the notion of 'protection of sense of self'. Unaffected siblings felt alien compared to their peers who did not have an ill sibling and chose to compartmentalise home life from school life in order to manage both worlds in a way that helped them to feel a sense of normality. The unaffected siblings felt they were responsible in protecting both their parents and their ill sibling. The theme of 'self as glue in relationships' described the multifaceted care that unaffected siblings gave towards their ill sibling and their parents. Some would choose to keep negative feelings or concerns to themselves in order to protect their family; acknowledging the stress that all members of the family were under and wishing to avoid contributing to this. 'Positioning in the adult world' emphasised the care duties that siblings took on, which for some would involve personal care duties. On the whole, this naturally evolved into care roles that were viewed positively through their being able to offer help and support to their siblings. Yet at the same time, this distanced them from shared experiences between themselves and their peers.

Brennan et al.'s (2013) study provided understanding of sibling experiences, which is often overlooked. The participants were recruited from a hospice setting where the majority of the children with the LLC had attended for less than 5 years. Some had been attending the hospice for over 10 years, showing a longer illness trajectory. It would have been beneficial for this study to separate these experiences in terms of length of illness trajectory. Combining these experiences together using grounded theory means that we cannot appreciate the element of time and the nuances of how temporality impacts upon the caring role.

A way of supporting siblings of children with a LLC was discussed in a review by Lane and Mason (2014) who also outlines and discusses a group intervention offered by a community children's palliative care service for siblings of children with a LLC. They suggested that siblings meeting other siblings who were in a similar position to them might have been helpful in reducing the sense of isolation and alienation that they felt due to the lack of shared experiences with their peers akin to results found above in Brennan et al.'s (2013) study. The group intervention intended to provide a safe environment where understanding of the illness experience can be spoken about; where they could express their opinions of how they felt about having a sibling who was unwell and where they could share coping strategies.

Lane and Mason (2014) outlined that in an evaluation of the group, siblings commented that they had enjoyed meeting other children in similar situations and had positive feedback overall for the HCPs who led the group. Having similar services available may help to reduce anxiety that unaffected siblings may feel. However, when considering Lane and Mason's (2014) review, much of the literature that they cited was taken from studies looking at siblings of children with intellectual disabilities or special needs. This should not be generalised to those living with a LLC. Intellectual disabilities would not be categorised as a LLC in the definition that is generally accepted and the care needs of these children would be different which would suggest a similarly different experience for siblings.

Adolescent caregiving for parents living with Huntington's disease (HD) was described as being a non-negotiable transition. Williams et al. (2009) conducted focus groups over a three year period with the intention of gaining understanding of care giving responsibilities, from the perspective of teenagers who had a parent with HD. Participants were all living with a family member who had tested positive for HD and some were displaying clinical symptoms. The results of this study indicated that the teenage caregivers displayed maturity beyond their years and expressed feelings of role reversal, where the teen felt they had swapped position to now be the parent. Although the participants seemed to manage with the caregiving tasks, emotional distress was apparent, with some disclosing that they were diagnosed with depression. An impact on relationships was found with caregiving restricting their social life whilst others expressed a sense of disengagement with society, which may be due to a lack of understanding of HD from their peers.

The use of focus groups as a method of understanding illness experiences raises some questions. Participants can build on the contribution from someone else's response, which could assist in clearly articulating their thoughts and being encouraged by shared experience. On the other hand, this could manipulate or hinder what the participant wants to convey and some may feel discouraged or self-conscious because they have different experiences to the other people in the group. Alternatively, by conducting additional one-to-one interviews, novel, more intimate data may have been generated.

This review will now move onto looking at social support more widely and the implications that living with illness has on these support systems.

2.2.3 The community implications of living with a LLC

The ramifications of living with a LLC have been shown to impinge upon the social life of the person diagnosed and their family. The complex care needs of a person may increase isolation from others for both the person diagnosed and their families (Runswick-Cole, 2010). One way this happens is the physical limitations associated with the disease progression, which can impact upon mobility and in turn be restrictive. This can be especially difficult if the person has been diagnosed later on in life when relationships have been established (Brewer et al., 2008; Lane & Mason, 2014). Jordan et al. (2015) found that parents of children diagnosed with a LLC felt disconnected and marginalised from society. This separation was even felt in *close* relationships with their family and friends due to the associated reduction in contact. Relationships and support are very important for people living with a LLC and the health of these individuals can in some ways be reliant on their relationships within their social structure (Cattell, 2001).

Gaveras, Kristiansen, Worth, Irshad and Sheikh (2014) explored what constituted social support for South Asian Muslim parents who were diagnosed with a LLC. They suggested that the support that is needed when living with a LLC was shown in both practical and emotional dimensions. Social support can be practical such as providing assistance with transport or offering personal care at home. It can also consist of offering emotional support such as being listened to and consoled. Lewis, Digiacomo, Luckett, Davidson and Currow (2013) found that culturally diverse groups and lower socioeconomic groups were less likely to participate within their community and were therefore less likely to receive help, whether from support groups or within the general population.

Concerns have been expressed as to whether these more vulnerable populations, such as ethnic minority groups in the UK, make use of supportive care services (Gaveras et al., 2014). One contributing factor previously outlined in the literature is that such groups allegedly do not need these services due to strong family support structures. This stereotype has been refuted in that migration has meant that it is more likely for extended family to live in different cities or countries (Gaveras et al., 2014). Yet, the association of ethnic minority groups with a strong support network has still been found to be held by HCPs, thus exacerbating the lack of referrals to supportive services, such as palliative care (Gaveras et al., 2014; Owens & Randhawa, 2004). In addition, Gaveras et al. (2014) found that parents diagnosed with a LLC described the unavailability of extended family members, suggesting a shift towards a nuclear family structure and highlighting the importance of enabling community services for this population. They found parents were also open to having both practical and emotional help from support services. Difficulties in cultural understanding increased the barriers to attending support services. Participants commented that some HCPs lacked appreciation for patients not being able to attend therapy or other medical appointments when it was at times of religious observance, such as Ramadan. This emphasises the need for palliative care services to take into account cultural differences, making sure that services are accessible for this population.

Lewis et al. (2013) argued that in order to better support people living with a LLC, community services should seek to extend their focus on a macro structural level; going beyond health service development by identifying resources that enable communities to work closer together. Lewis et al. (2013) suggested that creating and developing a community that is responsive to the needs of people living with a LLC in order to ensure better sustainability of resources is important. They conclude that focus needs to be given to support positive and beneficial facilitation of care, ensuring that more vulnerable populations are aware of the support that is available to them.

There is a paucity of research looking at the wider needs of people impacted by a LLC and whether community help or support groups could be beneficial. From the findings above, living with illness shakes one’s place within the world and causes feelings of segregation. Sontag (1983) described being a resident of either the kingdom of the well or a kingdom of the sick, reinforcing the alienation that characterises the illness experience. Support, both formal and informal, needs to come from a place of shared understanding of the vulnerability of the illness experience. The shared nature of being with illness needs to be further unpacked from an ontological perspective in order to better understand the nature of people’s experiences as deeply embedded in a relational world. Understanding more about these interdependent and subjective experiences, could illuminate from a basis of lived experience where care provision should support people on a macro-structural level.

2.2.4 Experiences with healthcare services

This section will now build on the prior focus on the individual, family and community experiences by incorporating research concerning the relationships between people living with an LLC, their families and HCPs. A LLC illness journey means significant interactions with HCPs (Johnston et al., 2016a). Wood, Simpson, Barnes and Hain (2010) interviewed parents of children with a LLC and found that parents complained of the inconsistency of healthcare staff, especially those concerning rehabilitation, such as physiotherapists and dieticians. Services that offered continuity through seeing the same consultant and other HCPs were praised by parents for their quality, demonstrating the importance of establishing relationships (Abbott, Carpenter, & Bushby, 2012).

Within the research base of experiences with HCPs, a strong emphasis has been given to the transition from being in children's palliative services to adult services. Adolescents and young adults are at risk of receiving disjointed care from healthcare providers as they fall between the categorisation of both child and adult care (Keim-Malpass & Lindley, 2017). The transition from childhood to adulthood has been shown to be related to modifications in relationships, roles and identity as well as a change in terms of desiring independence and autonomy (Kirk & Fraser, 2014). Fraser et al. (2014) estimated that over the previous decade, the number of 16 to 19 year olds with a LLC in England increased by almost 45%. The Marie Curie Trust (2012) suggested that under a quarter of children and adolescents who were being supported by children's hospices were now more than 16 years old (Fraser et al., 2014). This figure highlights the growing need to care appropriately for young adults who are transitioning from children's to adult services. If the condition was rare or often only specific to childhood or idiosyncratically different to those of older adults, adult services may not offer appropriate care for when young adults transition into their services (Ling, Payne, Connaire, & Mccarron, 2016; Rogers et al., 2011).

One difference found between the two categories of palliative services is the inclusion and provision of respite care for children compared to adults. Mitchell, Knighting, O'Brien and Jack (2016) found young people who used hospices for respite breaks found it to be very helpful for families in reducing stress, offering a break from the complex care demands. Young people also found children's hospices to be a supportive setting where they could discuss concerns with staff and feel listened to. Mitchell et al. (2016) found young adults and families were concerned and reluctant about the care offered in adult services and whether this would be appropriate for young adults. Although this study provides an insight into the experiences of hospice care from both the person with the LLC and their family, the people taking part were all white British, which limits the diversity in our understanding of this transition.

Transitioning from children's to adult services could be a positive and exciting period enabling new opportunities (Beresford & Stuttard, 2014; Martin House children's hospice, 2013) However, it could also be daunting and unwelcome, as young adults move from something that might be comfortable and safe to something that is unknown and different. Kirk and Fraser (2014) interviewed young people living with a LLC along with their parents and suggested additional difficulties in the transition between children's and adult services were the loss of long-standing relationships with HCPs in children's services, as well as the difference in organisation and culture between both services. Most parents described the transition between child and adult services as traumatic, with HCPs not being helpful in directing the transition in a clear and informed way. Little information was passed on to parents and they felt that a lack of care was affecting their child's health. Care was focused on the treatment of symptoms rather than offering a holistic approach that took into consideration the wider impact of the illness experience, leaving those concerned more vulnerable at an already highly emotional time. This lack of sensitive care presents the failure in the healthcare system to provide humanised care that ensures the whole person is treated rather than simply the condition (Galvin et al., 2018).

The studies reviewed above have recruited participants from hospice settings. More research is needed in the transition from child to adult services within a hospital setting. One such study by Beresford and Stuttard (2014) interviewed young people and their families living with a LLC and found that the young adults who were admitted into hospital unplanned due to infection or other causes experienced difficulty in the transition from children's to adult services. Parents were excluded from consultations and decision-making regarding care, and the young adults expressed feelings of social isolation. Additionally, parents commented on the lack of knowledge shown by healthcare staff of their child's LLC, as well as not recognising deterioration in their child's health.

The results of Beresford and Stuttard (2014) study reject the age-based dichotomy of care in favour of a more individual, patient-centred approach. From the narratives above, we can glimpse the concerns attached to care provision for people living with a LLC. Overall, from the research conducted, there is an under appreciation of the nuances of care for people living with a LLC. Taking a methodological approach that emphasises the idiographic, lived experiences of interactions with HCPs would illuminate more of the patients' needs and values, a factor of particular importance when considering the vulnerable nature of those living and dying with an incurable, progressive illness.

This section of the review has highlighted the impact and the meanings that living with a LLC has on the person diagnosed, their families and the ways in which illness transcends relationships and wider aspects of society. It has found that uncertainty and fear surrounding the illness trajectory is experienced. Relationships have been found to break down and community support groups can be non-existent or difficult to access. The experience of healthcare has been shown to be one of transition: transitions into different services due to age, or transitions between healthcare staff as continuity breaks down, causing a depersonalised approach to care. The next part of this review will focus on policy and care provision for those living with LLCs. The results of this part of the review of the literature will be used to critically evaluate policies and to help to illuminate the aspects of care that are missing or could be re-evaluated.

2.3 Definitions of Care

Due to the wide spectrum of diseases that qualify as being life-limiting, a person will follow their own distinctive illness trajectory. Some will require life-long medication and may need to have invasive medical procedures to help control symptoms, relieve pain and slow disease progression (Hain, Heckford, & McCulloch, 2012). Levels of care that are needed when diagnosed with a LLC can be complex until death or moderate and incremental due to a slow disease trajectory. Incorporated within any specific care given to treat symptoms of the LLC, palliative care and end-of-life care should be offered by HCPs as care pathways for people who are living with and dying of an incurable illness. The terms ‘palliative care’ and ‘end-of-life care’ are often used interchangeably in research, which can lead to confusion as to what each means. These varied definitions will now be explored using guidelines from best practice.

2.3.1 Palliative care

As a result of wanting to move away from religious motivations in caring for the dying in hospices, the WHO recognised palliative care as a major global health challenge (Clark, 2019) and formally defined it as being:

> *“The active total care of patients whose disease is not responsive to curative treatment. Control of pain, of other symptoms, and of psychological, social and spiritual problems, is paramount. The goal of palliative care is achievement of the best quality of life for patients and their families.” (The WHO, 1990).*

In 2002, the WHO altered their definition of palliative care to provide greater clarity and access at an earlier stage of being diagnosed with a LLC. This altered the focus of palliative care to be that which should be implemented across the illness trajectory as opposed to just at the end of a person’s life. Thus palliative care could be given even if the person has a long-term prognosis. The modified definition was:

> *“An approach that improves the quality of life of patients and their families facing the problem associated with life-threatening illness, through the prevention and relief of suffering by means of early identification and impeccable assessment and treatment of pain and other problems, physical, psychosocial and spiritual.” (WHO, 2002).*

Palliative care began to include specialisms in more areas of medicine such as, renal care, neurology and cardiology, as opposed to a pure focus on oncology (Clark, 2019). It is also characterised as being responsive to all ages. The WHO (1998) provided a separate definition of palliative care for children:

> *"Palliative care for children is the active total care of the child's body, mind and spirit, and also involves giving support to the family. It begins when illness is diagnosed, and continues regardless of whether or not a child receives treatment directed at the disease."*

Perhaps one of the most unappreciated features of palliative care is that it can operate in conjunction with other treatments that are given to prolong life such as chemotherapy or radiotherapy (Marie Curie, 2018). Thus the intention of palliative care is not to replace treatment, which is often a misconception even in HCPs (Gawande, 2010), but to work alongside it. Palliative care seeks to offer a holistic approach to care, as well as providing support for the whole family (ACT, 2011). The WHO (2002) guidelines state palliative care should:

- Affirm life and regard dying as a normal process;
- Intend neither to hasten or postpone death;
- Offer a support system to help patients live as actively as possibly until death;
- Offers a support system to help the family cope during the patient's illness and in their own bereavement;
- Use a team approach to address the needs of patients and their families, including bereavement counselling, if indicated;
- Enhance QoL, and may also positively influence the course of illness.

More recently, a debate has arisen as to whether palliative care should be re-defined (Boyd, Moine, Murray, Bowman, & Brun, 2019; Clark, 2019). The International Association of Hospice and Palliative Care (2018) suggested that the definition of palliative care should be:

> *"Palliative care is the active holistic care of individuals across all ages with serious health-related suffering due to severe illness, and especially of those near the end of life. It aims to improve the quality of life of patients, their families and their caregivers."*

Examples of further qualifiers that were provided as to what palliative care should encompass are as follows:

- Includes prevention, early identification, comprehensive assessment and management of physical issues, including pain and other distressing symptoms, psychological distress, spiritual distress and social needs. Whenever possible, these interventions must be evidence based.
- Provides support to help patients live as fully as possible until death by facilitating effective communication, helping them and their families determine goals of care.
- Is applicable throughout the course of an illness, according to the patient's needs.
- Is provided in conjunction with disease modifying therapies whenever needed.
- Is applicable throughout all health care settings (place of residence and institutions) and in all levels (primary to tertiary).
- Can be provided by professionals with basic palliative care training.
- Requires specialist palliative care with a multiprofessional team for referral of complex cases.

The International Association of Hospice and Palliative Care (2018) further suggested that in order to integrate palliative care more effectively, governments should:

1. Adopt adequate policies and norms that include palliative care in health laws, national health programmes and national health budgets;
2. Ensure that insurance plans integrate palliative care as a component of programs;
3. Ensure access to essential medicines and technologies for pain relief and palliative care, including paediatric formulations;
4. Ensure that palliative care is part of all health services (from community health-based programs to hospitals), that everyone is assessed, and that all staff can provide basic palliative care with specialist teams available for referral and consultation;
5. Ensure access to adequate palliative care for vulnerable groups, including children and older persons;
6. Engage with universities, academia and teaching hospitals to include palliative care research as well as palliative care training as an integral component of ongoing education, including basic, intermediate, specialist, and continuing education.

This definition, although similar in many regards to the WHO's (2002) description, intends to add more detail and emphasis on palliative care being available from 'early identification' of a LLC. It also highlights the importance of training healthcare providers in palliative care and developing specialists in this remit for complex care cases. Clark (2019) criticises the notion of having a universal definition of palliative care arguing:

> *"culture, wealth, poverty, the presence or absence of universal health coverage, corruption, levels of health system delivery, and a myriad of other factors will shape how palliative care is perceived, delivered and defined in any given situation."* (para. 24).

It is also clear from the guidelines that they envision a significant role for governments in raising awareness of and noramalising good practice in palliative care. This kind of leadership would allow for the development of the kind of macro structural level implementation that Lewis et al. (2013) advocated above to protect vulnerable groups.

In the UK, the NHS (2018) defines palliative care as a conduit for end-of-life care and that its intentions are to make the person with the LLC feel as comfortable as possible throughout their illness, by managing pain and other symptoms attuned to the holistic approaches defined above. The terminal illness charity, Marie Curie (2018) defines palliative care as "treatment, care and support for people with a life-limiting illness" and that palliative care can sometimes be called 'supportive care', which suggests a purposeful movement away from the connotations that are attached to palliative care as care only when death is imminent (para 2). Bowden et al. (2018) commented that the terms 'best supportive care' and 'palliative care' are often interchangeable but can take on the same meaning within the specifics of a clinical context. They claim that the de facto realisation of 'best supportive care' for patients is not a meaningful definition (Bowden et al., 2018).

In the UK, palliative care is provided by a variety of institutions including the public, private and voluntary sectors (Hain, Devins, Hastings, & Noyes, 2013). Palliative care can be received in a number of settings: in primary and tertiary care facilities, in community health centres, at home, hospitals or hospices (Marie Curie, 2018). Guidelines highlight that palliative care is multidisciplinary in its approach, including the 'parent/carer' (referring to spouse, partner or next of kin) in decision-making. Specialist palliative care services can be called on if required, which have consultants trained in palliative medicine, specialist palliative care nurses and specialist occupational therapists or physiotherapists (NHS, 2018). The NHS (2018) outlines that if a person is being cared for at home, the GP has overall responsibility for the person's care.

2.3.2 End-of-life care

The NICE (2017b) guidelines recommend that end-of-life care should be managed as a long-term process, beginning at the time of diagnosis and developed through plans for the person's future in terms of their overall care, including care related to the dying process. It is therefore similar to the WHO's (2002) definition of *palliative care.* The NICE (2017b) guidelines differentiate 'end-of-life care' from the status of 'approaching the end of life', which describes the phase of illness after a change in the person's condition that means they are likely to die within weeks. In contrast, the General Medical Council (2010) define approaching the end-of-life as a person who is likely to die in the next 12 months. It is unclear from the NICE (2017b) definition as to the difference between palliative care and end-of-life care. The NICE (2017b) definition seemingly includes palliative care within end-of-life care, akin to the NHS (2018) guidelines on end-of-life that include palliative care in the definition. Therefore definitions of palliative care and end-of-life care are not distinct.

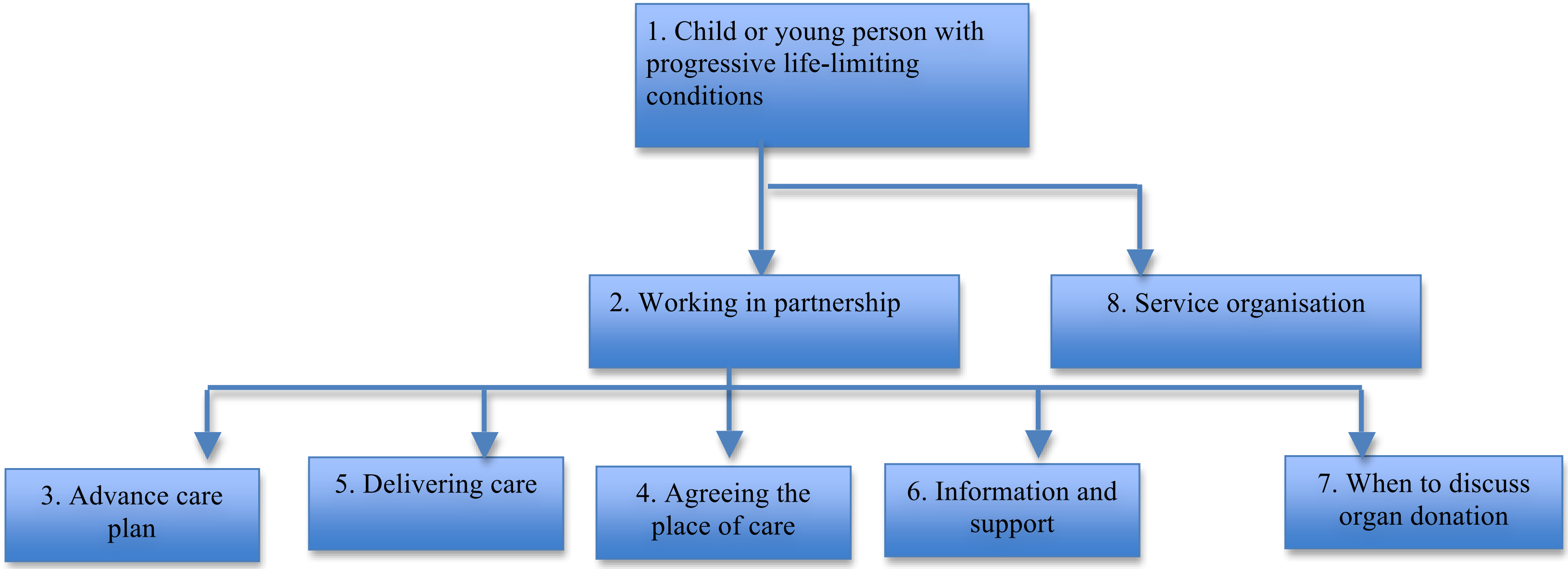
The NICE guidelines for children and young people living with a LLC were commissioned with the purpose of standardising care that is consistent and available to all (Villanueva, Murphy, Vickers, Harrop, & Dworzynski, 2016). In their "planning and managing end of life care for a child or young person with a life-limiting condition" guidelines, NICE (2017b) provide a care pathway diagram defining the direction in which care should be managed (see Figure 1). The contents in this figure seem to advocate the importance of patient-centred care, good communication and a multidisciplinary care approach. Similar to the WHO's guidelines, NICE (2017b) recognise end-of-life care as a partnership between the patient and their parent/carer in that they are important participants in the decision-making and care planning process. Members of a patient's multidisciplinary care team should be known and their roles should be explained to the person with the LLC and their parent/carer. The NICE (2017b) guidelines state that each person diagnosed with a LLC should have a medical lead specialist who coordinates care and that this should be explained to the person and their parent/carer. Like the qualifiers in the WHO's (2002) definition of palliative care, NICE (2017b) stipulate that the care team should consist of primary, secondary or tertiary services, including any specialists in the underlying LLC, hospice professionals and members of the specialist palliative care team, if needed. Allied health professionals as well as social care practitioners and chaplains should be involved with care to meet with offering a holistic approach.

NICE (2017a) provide a separate set of guidelines for HCPs to adhere to when people living with a LLC are in the last few days of life. These guidelines include the following as being the most important considerations:

1. Hydration and nutrition;
2. Managing physical distress and discomfort;
3. Symptoms and signs of imminent death;
4. Transfer to preferred place of death.

Thus, these definitions of end-of-life care and approaching end-of-life encompass a broad and sometimes contradictory spectrum of care for those diagnosed with a LLC. The confusion between the terms will be discussed next.

Figure 1. The NICE (2017b) flowchart of planning and managing end of life care for a child of young person with a life-limiting condition.



2.3.3 Analysis of care definitions

The complexity of the illness trajectory, the difficulty of navigating the point in illness when further end-of-life care is considered and the experience of the last few days of life calls for variation in guidelines to enable HCPs to care sensitively and appropriately. However, the interchangeable use of both palliative care and end-of-life care listed above causes confusion surrounding the meaning of these types of care and impacts upon the decision-making of whether to accept these care pathways. Boyd et al. (2019) commented: "Nearly 30 years after the World Health Organisation first defined 'palliative care', we are no closer to agreeing what the term means." (p.1).

An example of this confusion can be seen in Marie Curie's (2018) definition stating that "having palliative care doesn't necessarily mean that you're likely to die soon – some people receive palliative care for years [..] Palliative care is also for people nearing the end of life – this is sometimes called end of life care." (para 5). Marie Curie (2018) suggests that treatment and care given in end-of-life care "is an important part of palliative care." (para 7). The interchangeable use of these definitions, not just within key services in the UK but globally, causes confusion. The NICE (2017b) end-of-life care guidelines are similar to the guidelines that the WHO (2002) published but are instead referred to as palliative care.

The confusions surrounding definitions has a range of impacts upon the decision-making of stakeholders in the process. Firstly, the variation as to when palliative or end-of-life care is offered by HCPs depends upon their conceptualisation of the terms. Secondly, if the term 'end-of-life care' is discussed by HCPs with patients as a pathway, this could impact upon whether care is accepted by patients and their families due to the connotations that the phrase holds. The paradoxical conflation of palliative and end-of-life care exacerbates the problematic stigmatisation that dominates public perception of such care, thus reinforcing the decisions of both HCPs and patients when planning care (Bailey & Cogle, 2018). This impedes upon the benefits that such care can offer the person with the LLC and their families.

All definitions of palliative care and end-of-life care say that this care prioritises improvement to QoL. However, it is unclear in the definitions how improving QoL in people who are living with a LLC is operationalised in practice. This becomes particularly ambiguous when making decisions surrounding the best interest of the patient and withdrawing or withholding care. From the definitions above, it could be inferred that HCPs often have to make decisions based on their own intuition and experience (Monterosso, Ross-Adjie, Rogers, Shearer, & Rogers, 2016). Many of the elements within the definitions are dependent upon when the HCP deems it to be the 'right time'. For example, when is the right time to stop treatment and allow death to take its course? What does QoL look like in the last few days of life? This illuminates the complexity for HCPs working within palliative care and the difficult, life-altering decisions they have to make on a daily basis. The NICE (2017) guidelines on delivering care for a child or young person at the end of life, as well as the guidelines for adults, provide practical advice surrounding the physical bodily deterioration and objective signs that death is soon approaching, such as changes to breathing and reduced consciousness, but decisions surrounding stopping treatment is mainly dependent upon clinical judgment.

Seeking to remove the stigmas around palliative care that are currently exaggerated by conflated definitions would necessarily lead to more people participating within such care from an earlier point in their illness. This would engender a particular set of practical considerations. An analysis conducted by Marie Curie reported that the NHS is struggling to cope with end-of-life emergency admissions, particularly in England, which if it continues would mean an estimated cost to the NHS potentially doubling to £4.9bn by 2041, with over 8000 extra hospital beds needed (Thornton, 2018). These admissions could be avoided if sufficient care within the community was provided, thus suggesting practical and fiscal benefits to addressing these concerns (Thornton, 2018). Securing good palliative care across the illness experience may aid in avoiding an emergency admission into hospital towards the end-of-life, an experience that can be stressful and uncomfortable in unfamiliar surroundings. Of course, it should also be recognised that catering for palliative care for all people diagnosed with a LLC at the beginning of their illness experience means that a large number of patients and professionals will be involved and the strains that this will have on services need to be considered.

Perhaps it could be argued that there is a deeper underlying reason as to why there is a seemingly deliberate obfuscation of palliative and end-of-life care definitions and pathways. This is rooted in the taboo in western culture of talking about death and dying. Arguably as a society, we are not taking part in palliative care due to the necessary acknowledgement of death that results from such engagement. The suggestion that redefining palliative care as 'supportive care' or adding the words 'early' or 'integrated' into its label would encourage people to take part in palliative care from diagnosis (Boyd et al., 2019) is based on the emphasis that death is still far in the future, attempting to disassociate palliative care from what it is actually for, namely those who are dying. Altering the language of palliative and end-of-life care may serve to encourage more of those living with a LLC to participate and to see it as separate to care provided when one is dying, but careful consideration needs to be given to the potential consequences of such a change. The potential for unintended consequences in divorcing such care from the notion of death may only compound the issue of our inability to face our mortality, thus failing to do any more than paper over the cracks in care provision for those most in need. The next sections will look at care provision for these types of care and the utilisation of these in clinical practice. Through synthesising the literature this will concentrate on: models to aid communication, training for HCPs and decision-making in end-of-life care.

2.4 Care provision and effective communication

Within healthcare policy there has been a movement away from a paternalistic approach to care to one that is inclusive of the patient and their families as a central priority shown in The King's Fund report (Ham, Berwick, & Dixon, 2016; Walker & Waterworth, 2017). One way of facilitating patient-centred care when people are living with a LLC is the Advanced Care Plan (ACP). An ACP is a formal document that records the individual's wishes and preferences for care when living with a LLC, providing a sense of autonomy (Compassion in dying, n.d.; NHS, 2018). The NICE (2017a) define an ACP as:

> *"The process of discussing your preferences and wishes about future treatment and care with those close to you and your healthcare team. The process may include talking about where you want to be cared for, identifying the people you would like to be consulted about your care, or making treatment decisions in advance." (p.1).*

Originally, communication of the patient's preferences was conducted at the end-of-life (Barnes et al., 2012) but over time ACPs have been developed to cover a broader range of issues to encourage engagement in conversations related to the values, fears and hopes of the person with a LLC and their caregivers (Pearce & Ridley, 2016). The ACP should be shared within the patient's multidisciplinary team and the document should also encourage patients to have a named proxy in place in case they are no longer able to make decisions due to cognitive decline (Pearce & Ridley, 2016). ACPs can be used from the beginning of the illness trajectory, yet disagreement as to the timing of such conversations has been observed amongst clinicians in that it is best to avoid premature conversation in order to reduce the risk of patient distress (Pearce & Ridley, 2016). Initiating discussions around ACPs can be challenging especially if the condition is stable as conversations around wishes can seem untimely (Mitchell & Dale, 2015). However, early development of ACPs can help in aiding care if the person with a LLC unexpectedly deteriorates. Mitchell and Dale (2015) found that critically unwell children who were admitted into hospital without having a prior chance to complete an ACP faced challenges due to not having time to prepare or think about treatment preferences with their team of HCPs.

Hiscock and Barclay (2017) interviewed a variety of HCPs, including respiratory physicians and nurses, community paediatricians and hospice family support workers to discuss the most appropriate times to have conversations with their patients about ACPs. They found that the most appropriate time to start the discussion according to the participants seemed to be when the disease had progressed, but the patient was not in end-of-life stages. However, timing this discussion was described as being difficult due to the uncertainty in illness trajectory. Pearce and Ridley (2016) argued that it was important for HCPs to initiate an ACP discussion by normalising the conversation as one which is beneficial to the patient and something that all HCPs do. However, as they also point out, it is vital for the HCP to perceive how their patient understands their own health, as this will affect the direction in which the conversation about the ACP goes. If a person or their family has not accepted their illness prognosis, or believes that their health will improve, discussion about an ACP may be something that causes frustration and distress (Bluebond-Langner, Hargrave, Henderson, & Langner, 2017). Thus the timeliness of initiating these conversations is of great importance to give space to reflecting on the finality of life and how people would like to spend their remaining time. In addition, continuity of care whereby HCPs have been able to build a relationship with their patients is beneficial in discerning an appropriate time to broach these conversations.

Mitchell and Dale (2015) investigated current practices in ACPs by interviewing consultants and senior nurses who have experience of conducting ACPs for paediatric patients. Similar to Pearce and Ridley (2016), Mitchell and Dale (2015) found that the uncertainty in illness trajectory in LLCs created a barrier to communicating about and creating an ACP. However, all participants thought ACPs were essential and beneficial. They found that nurses in their study could recall instances where an ACP discussion had resulted in the achievement of a peaceful terminal phase of illness and death in a preferred place of care, which is essentially the key outcome of ACPs. An ACP can also reduce the chance of an aggressive experience of care at the end of life, due to decisions being made about treatment when it is no longer appropriate. Conversely, Mitchell and Dale (2015) found deaths were described in which ACPs had been lacking or inadequate, resulting in what was believed to have been substantial distress for the patient, the family and the staff involved. The reality that these problems persist serves to advocate the benefits of ACPs, but in a way that is sensitive to the beliefs and values of the person and their family and their acceptance of their illness trajectory.

In spite of the benefits attached to ACPs, the ‘End of Life Care audit: dying in hospital’, for England 2016 showed that only 4% of patients had documented evidence of an ACP. In a report for the Royal College of Physicians, Bailey and Cogle (2018) outlined that the results of this audit could be due to: a failure from HCPs in completing an ACP; a lack of documentation of these wishes; or the failure to share these documents between services. The challenges attached to recording ACP conversations and sharing them appropriately across services, which use different IT systems, is a barrier. Other barriers to not completing an ACP have also been found, including a lack of time for HCPs to discuss such difficult topics, as well as not having enough time to build a rapport capable of broaching this with families (Mitchell & Dale, 2015).

Hiscock and Barclay (2017) found that there was no set pattern as to when HCPs would begin a conversation about an ACP and that each individual was different, so a case-by-case decision was made built on the HCP's judgement. Bailey and Cogle (2018) argued that throughout a person's illness experience, there are multiple opportunities for HCPs to have a conversation with patients surrounding plans of treatment and their preferences towards the end of their life including location of death. Yet, in practice, it appears that some HCPs doubt that they are the 'correct' person to be having such conversations with the patient and instead defer to other clinicians. For example, the person's GP may feel that the specialist would be more appropriate in discussing palliative care, yet the specialist may feel the GP knows the person best and so should instigate the conversation (Bailey & Cogle, 2018). However, increasingly, people see whichever GP is available at the time rather than waiting to see their designated GP, thus the nature of the relationship between patient and their GP is questionable in the context of current practice.

The reticence of both HCPs and patients to discuss issues surrounding ACPs can again be attributed to the taboo of death that remains in Western culture; especially the death of a child. Dying and death have become uncomfortable and 'morbid' to talk about, and thus resisted or denied (Zimmermann & Rodin, 2004). Hancock et al. (2007) conducted a systematic review of 'truth-telling', i.e. the degree of honest communication about the person's prognosis in HCPs who were caring for people with a LLC. Hancock et al. (2007) included 46 articles and found that HCPs withheld information or were reluctant to discuss prognosis and end of life issues with their patients for a number of reasons. These included:

1. Discomfort of HCPs in disclosing information especially in terms of 'do not resuscitate' orders;

2. Uncertainty about illness trajectory;

3. Time constraints in consultation;

4. Patient requests for information - three included studies found that if the patient did not request information, the doctor would not disclose their prognosis;

5. Concerns for negative impact on patient - some included studies found that doctors would not reveal prognosis details if they felt that the patient could not manage the information. HCPs thought it was best that the patient still had hope in the most ethical way that they could, which perhaps would not be present if they disclosed their prognosis;

6. Family/caregiver requests for withholding of information - asking the HCP to withhold information concerning the patient as families felt that it was in the best interest for the person with a LLC to not know, even though they were adults. However, in two included studies, HCPs said that the patient's autonomy and information preferences were paramount.

Research such as this demonstrates the complexity of caring for people with incurable illness and the ambiguity surrounding when and how to have discussions of these matters. With consultations with GPs only being around ten minutes in duration (Iacobucci, 2018), the busyness of the outpatient clinics in hospitals, the lack of privacy on a hospital ward or the uncomfortable surroundings in a hospital cubicle when a patient is bed bound makes it difficult to hold these conversations in a sensitive manner. The Royal College of Physicians (2018) report that often these conversations are held at crisis points, in emergency settings, when people are unprepared and clinicians are not known to patients and their families (Bailey & Cogle, 2018).

A framework that recognises the importance of these conversations is the 'second conversation' model (2019), which is currently being piloted at three NHS trusts in London ("The second conversation: Improving training around end-of-life care conversations | RCP London," n.d.). The rationale behind developing the model for people living with a LLC comes from clinical practice where end-of-life care conversations are often led by a senior clinician, either a consultant or a specialist registrar. The conversation centres on care plans regarding the withdrawing or the withholding of treatment (Bailey & Cogle, 2018). These conversations are challenging in that patients could feel like they are being abandoned or dismissed. The challenge of initiating these conversations from a HCP's perspective can mean that they are not held, or are conducted insensitively. The potential to rush could deny patients the opportunity to reflect on what is important to them with regards to care at the end of their life.

Best practice guidelines suggest that these initial conversations should be followed up in order to ensure that patients have the chance to ask questions and also to communicate any other wishes, having had time to reflect on their care. The ‘second conversation’ model proposes that these follow-up conversations should be led by a foundation trainee medic, in order for them to build their skills and confidence in these discussions of end-of-life care. The aim is that this would serve as an educational tool in breaking down the barriers for early career medics in talking about death and dying. One reason why HCPs, especially junior doctors, may find it difficult to talk through end-of-life issues is due to the perspective that they bring to bear on the situation. Carel (2017) conceptualised the notion of the outsider (third-person) perspective vs. the insider (first-person) perspective. Carel (2017) concluded from a review of literature that the outsider who does not have an illness perceives a LLC as being worse compared to the perspective of the person who is ill. She suggested that healthy people have a vague and imprecise idea of what living with an illness entails, especially when the disease may be rare and fewer people know about it. The outsider may perceive that the ill person’s life is being defined by illness. As a result there may be a failure of the outsider to adopt a view that finds synchronicity with the person who is ill and their perspective of disease progression. This could be true of HCPs too; although they care for people who are ill on a daily basis, they may perceive the person as not being able to manage conversations regarding end-of-life care or feelings towards death.

Carel’s (2017) perspective should be appreciated more within a clinical setting to ensure HCPs do not make assumptions as to how the person may respond when discussing end-of-life care. Models like the ‘second conversation’ is one initiative to enable junior doctors to experience these conversations more, surrounding themselves with talking about death rather than concentrating on medical intervention.

Language use is crucial when HCPs are communicating with people who have a LLC and their parent/carer. The particular phraseology associated with palliative care can have potentially harmful consequences for the patient and their parent/carer (Boyd et al., 2019) and create negative connotations towards such care. When HCPs are discussing treatment options or indeed if treatment is no longer working, language such as ‘withholding treatment’ or ‘treatment withdrawal’ can impact greatly on the patient and family’s understanding of the care that they are receiving, and may lead to feelings of neglect. The term ‘ceiling of care’ is used within recommendations for clinicians (Bailey & Cogle, 2018; Boyd et al., 2019). This term is far removed from a sensitive and humanised approach to communicating about this profound and existential moment in a person’s life. This could impact greatly upon the patient’s relationship to end-of-life and their acceptance of death. The challenge for HCPs is to communicate with patients in a way that is empathic and relational but in a way that acknowledges the certainty of death. To do so would be to counteract the fact that an honest relationship with mortality has been overshadowed by the medicalised society in which we live, where science is seen to owe us a long and healthy life.

2.5 Policy in practice and training for healthcare professionals working in palliative and end-of-life care

The WHO (2002) suggests that palliative care can be positively implemented in spite of limited resources. This is questionable as studies have demonstrated the lack of awareness that HCPs have in relation to their conceptualisation and operationalisation of palliative care. One reason for this is the limited training in palliative and end-of-life care HCPs receive. Monterosso et al. (2016) conducted focus groups in order to explore HCPs' understanding of and attitudes towards palliative care within a multidisciplinary team including clinicians, nurses, midwives and providers of pastoral care. Monterosso et al. (2016) found that HCPs believed good palliative care incorporated keeping the patient pain-free and comfortable and making sure that the family was regularly informed of care treatment and options. Early incorporation of palliative care within the illness trajectory was viewed as favourable.

Continuing with describing palliative and end-of-life care in practice, Spalding and Yardley (2016) explored children's, parents' and hospice staff's opinions on what medical students should learn in preparation for their careers as doctors. Their rationale for doing this work was the lack of self-efficacy medical students have in paediatric palliative care compared with other specialities. Participants related their experiences with HCPs or patients and specified that which they found helpful or unhelpful within these interactions. Themes from the data indicated that: children like to be included in decision-making regarding their treatment, giving value to focusing on explaining things clearly so that children can understand what is being said; and parents placed a high value on doctors acting compassionately. Additionally, the third and most significant theme was that of interpersonal communication, both verbal and non-verbal, such as body language and touch, and how important it was to children and their families that doctors interact with them at their level using appropriate language. This approach would enable parents to connect more with HCPs in a way that is personable, thus helping in planning for their child's care needs. It also presents a need to enhance humane and sensitive practice, where patients along with their families feel care is relational as opposed to them feeling 'other' or 'objectified' (Galvin et al., 2018). Galvin (2010) suggested that leading care that is humanised is a balance between the:

Hand – technical skills

Head – protocols and evidence

Heart – ethical and human dimensions

This balance is especially relevant to the notion of phronesis, which is sometimes referred to as practical wisdom. In the context of this review, an example of practical wisdom would be related to HCPs' decision-making about when to talk about end-of-life care to a patient, when to withdraw or withhold care, and how to communicate this to a patient and their family. A hand-head-heart approach ensures that HCPs care for *people* living with LLCs not just "physiological systems" (Galvin, 2010: p.170).

Behind this approach is an understanding of the nature of being human. We do not just experience our bodies as "skin, muscle, bone or organs" (Toombs, 1993 cited in Galvin, 2010: p.170), but we are living, relational, experiential beings. This is no different when one is dying. As Galvin (2010) comments: "*even when critically ill, ventilated or unconscious, the human person's experience is not compartmentalized*" (p.169). Much focus within the medical world is placed on the *hand* element, the technical skills of HCPs, using the spirit of technology to understand the human *body* that they are treating, seeing the 'patient' through the lens of the objective gaze. Of course, the hand has a role to play but this is sometimes limited within end-of-life care where prolonging life is often not the primary goal. The limitations of some HCPs was found by Zambrano, Chur-Hansen and Crawford (2013) where surgeons who were caring for people with a LLC described their relationship style to their patients as detached and emotionally distant. Practically, this entailed not knowing many details about the patient and their personal situations in order to act more objectively (Zambrano et al., 2013). The juxtaposition of curative care and end-of-life care are seen here. Both are in the medical world yet are opposing ends of the spectrum. One tries to prevent death, whilst the other recognises it and wants it to be done in the best way for the person.

Reviewing the literature, it seems that training is crucial, but is missing when considering the areas of palliative and end-of-life care. HCPs who work within general hospital departments will sometimes care for people with a LLC and thus should receive training that will help them to manage the complexities that come with caring for these people. The multifaceted challenges that a person living with a LLC has on a daily basis has been found to include physiological, psychological, ethical and spiritual concerns (Stayer, 2012). Research has suggested that multidisciplinary staff who work with people with a LLC require further training, especially with regards to the issues surrounding existentialism and spirituality that people face when they are within a palliative care setting (Llewellyn et al., 2015; Walker & Waterworth, 2017; Weaver & Wratchford, 2017). Training should be considered especially crucial for nurses, as they have been found to spend a significant amount of time providing care for people and families with a LLC compared to other HCPs (Forster et al., 2017; Stayer, 2012). Stayer (2012) found that typically, paediatric nurses do not receive the same formal education training as hospice-based and palliative care nurses. Additionally, they do not have the same support systems in place, which offer a supervisory network whereby staff can have access to someone to talk to when affected by the situations that they are dealing with.

Similar results with regards to the lack of training for HCPs to care for people with LLCs were found in Forster et al's. (2017) study, which looked at trainee psychiatrists who cared for people who were at the end of their life, with one trainee calling the lack of education in this area "an educational famine" (p.233). They found that trainee psychiatrists felt ill-equipped to talk to people who were dying which caused them to feel anxious. Some trainees recalled only having two to three hours of training in issues at the end-of-life, whilst others were unable to recall having any training on this. From these results, trainees presumed that the more people they care for who have palliative care needs, the more they would know how to communicate with them. However, from other research, HCPs who have been working in the profession for a long time still felt ill-equipped to care for people within palliative care, especially in terms of communicating with this population (Morris et al., 2012).

In comparison to this, palliative care nurses who work within hospices receive formal education regarding the complexities of end-of-life care and death, as well as having formal support systems in place such as regular supervision to discuss experiences (Forster et al., 2017; Stayer, 2012). With this said, a recent report by Public Health England (2018) found more people still die in a hospital compared to a hospice, so therefore, all hospital staff who care for patients should undergo similar training and support. As Santos and Moreira (2014) state when interviewing HCPs who care for children and young people with LLCs, *"Health professionals that experience the living and dying process of children and adolescents…carry marks with them*" (p.4876), emphasising the difficult nature of caring for people in this position and the long-term effect that these experiences can have on HCPs' lives.

Ethical dilemmas are a central component of knowledge of end-of-life care in training staff, but focus should also be given to monitoring HCPs and the challenging cases of life and death that they are involved in each day. Galvin (2010) provides an example of a nurse caring for a dying patient. The nurse is not just a bystander, but shares in the lived world alongside the patient. This experience intertwines his own vulnerability with his own mortality, or his family's and his loved one's. The finality of life is seen first-hand as the nurse cares for the dying patient. The nurse's care is straddled between caring for the objective body and also the subjective vulnerability of the dying patient along with her family. Galvin and Todres (2009) argue that leading care from this recognition of both the objective body and the lived body can direct care to being holistic and humanised. Allowing an embodied understanding to direct care involves "a living access to important feelings and insights that are carried and affirmed and are more than just principles that are thought" (Galvin & Todres, 2009: p.147). This embodied understanding is the *heart* of care from the hand-head-heart approach (Galvin, 2010) previously mentioned. Thus, caring in a way that does not depersonalise the patient but is relational and sensitive to the shared meanings of body and world that is paramount. This response is fluid in its balance between the three unified ways of caring. Galvin and Todres (2009) label this care as 'openheartedness'. This human approach to caring is especially necessary within palliative and end-of-life care cases. Following more humanised-led care within palliative and end-of-life settings would not only enhance care received by patients and their families, but would also humanise the medical world for HCPs who would benefit from the opportunity to consider seriously the intrinsic emotional weight of their experiences.

A level of vulnerability was discussed in Robinson's (2019) perspective piece asking whether doctors should cry at work. Robinson (2019) discussed how some HCPs working within palliative care do not allow themselves to feel emotional with their dying patients as this could be seen as a weakness or unprofessional. A quote from an anaesthetic registrar revealed:

> *"We work in an environment full of emotion, and developing the skills to help us demonstrate our care, vulnerability and humanity in a sustainable and healthy way is essential – yet it's so rarely taught." (Robinson, 2019: p.2).*

The stringent policies that are in place, and that lead the care provision, have in some ways overshadowed the vulnerability of HCPs when considering good practice in the context of life and death situations. Some hospitals in the UK are now conducting 'Schwartz rounds'. This provides an informal yet structured forum, which is open for all staff, clinical and non-clinical staff, to come together regularly to discuss the emotional aspects of working in healthcare ("Schwartz Rounds | Point of Care Foundation," n.d.). This is a movement away from trying to 'solve' problems related to treatment, but instead serves as a reflective platform toward the intrinsic challenges that come with caring for others. This initiative was developed through a patient's witness to the compassionate care he received when he was dying from lung cancer. He said:

> *"I have learned that medicine is not merely about performing tests or surgeries, or administering drugs… For as skilled and knowledgeable as my caregivers are, what matters most is that they have empathised with me in a way that gives me hope and makes me feel like a human being, not just an illness." (Ken Schwartz as cited in The Point of Care Foundation, n.d.).*

These lived experiences of HCPs when caring for patients and their families are not just theoretical and should not be dismissed. Instead these experiences should be utilised to lead the informing of good practice within palliative and end-of-life care. Through reflective practices such as the Schwartz rounds, HCPs may feel better supported to then enable them to better support and care for their patients in turn. This care should recognise a shared sense of vulnerability, based on our primordial human connectedness in order to act in an "empathically sensitive actionable knowledge" (Galvin, 2010: p.173).

2.6 Timing the shift from living to dying

The timing of when to end curative or management treatments due to them no longer being useful for people living with a LLC is a difficult decision to make. Gawande (2016) argued that within the medical and research worlds, too much time is spent on how to prolong life in terms of quantity and less time is given to how to live with illness well. Gawande (2016) suggested that this continuation of using curative treatments and not stopping them at an appropriate time has been a foremost contributor to the underuse of hospices and the lack of recognition of the significance and value of engaging with palliative care. He argued that people who persist with curative treatment instead of accepting and moving into 'approaching end-of-life care' (NICE, 2017b) have a more painful and uncomfortable dying experience due to the persistence of clinical interventions, as seen in Mitchell and Dale's (2015) study on the use of ACPs above. The guidelines outlined do not clarify when to stop the curative treatments and transition into end-of-life care. The law states that withholding or withdrawing treatment, for example the withdrawal of hydration or nutrition, is allowed when the treatment is not in the best interest of the person with the illness. It is currently against the law in the UK to end life when requested (General Medical Council, 2010; NICE, 2017b; Wright et al., 2009). This is where differences of opinion can become more prominent and serious, especially between clinicians and the family members of the person who is ill, as the task of working out a plan is a delicate and complex process. Effective communication is essential from HCPs from diagnosis and throughout the illness journey, in order to come to a decision regarding care in the most agreeable way possible, where a rapport between both the families and the HCPs mean that there is a level of trust in making a combined decision.

Monterosso et al. (2016) found that some HCPs felt uncertain and uncomfortable about making decisions regarding the withholding of treatment when a patient was no longer responding to intervention. This illuminates the difficult decisions that some HCPs have to make and how these are formed using their own judgements. Continuing the hand-head-heart approach to care, Galvin and Todres (2009) suggest that the felt sense, which is the way in which meanings are implicit to our bodily experience, should be utilised as a resource to guide care that is separate from a set of deterministic principles. By applying felt sense to a care situation, HCPs would be guided on more "implicit existential and personal information that draws on what one has lived through" (Galvin & Todres, 2009: p.147). Thus this provides a way of care, which connects the personal with the professional, through an embodied understanding of the patient's experience. Galvin and Todres (2009) argue that in order for the felt sense to be used as a resource it needs to be conceived of as an embodied experience, something HCPs can relate to within their own body, rather than as an abstract concept; awareness of it should therefore be facilitated in training HCPs.

Zambrano et al. (2013) investigated surgeons' involvement with people who were living with a LLC and decisions around treatment. The complex questions of the benefits to a person who is nearing the end of their life with regards to how effective surgical procedures may be was explored. Through interviews, Zambrano et al. (2013) found that surgeons often identified themselves as someone who could fix and cure a *problem*. They found that within the surgery culture, dying and death are perceived as failures to themselves as surgeons and that death could be put on hold through operating. In some cases, it is questionable as to whether death being put on hold would be the right thing for the patient. Would they benefit from a longer life or would it in fact be painful and more debilitating? The medical curative world here meets the patient experience world and the weighing of these decisions adds to the complexity of what is best for the patient from the viewpoint of both worlds. In not wanting to accept failure, there is a sense of surgeons emphasising the 'otherness' of the patient, viewing the body as an object, a puzzle to solve and fix rather than as a relational being. Galvin (2010) argues that this sort of withdrawal which HCPs make is the easier and less pain-filled option to take when caring for a patient. Galvin (2010) describes it as taking "refuge in a voyeuristic stance of the fascination of 'this is not a person and it is not me in there'," (p.173) thus describing the creation of a distance between the shared meanings of being human, and the shared experience of dying we will all endure. As a result, the surgeon is in the world of the healthy and of ability where death equates to failure and so the personhood of the patient is rejected in favour of an objectified, meaning-free body that cannot interfere with this priority. A HCP with this mind-set embedded in their professional identity may encounter significant challenges in navigating the transition from curative to end-of-life care, a realm of experience for which their practice has left them ill-equipped.

This medicalised view of the world can be seen as a possible consequence of an industrialised and marketised care outlook where competition is the driver of excellence. It is therefore normal practice to intervene to save a life, even if the repercussions which come from this are overtly harmful, such as additional infections from a gastrostomy tube or broken ribs from conducting Cardiopulmonary Resucitation (CPR) (O'Mahony, 2016). Having said this, The Royal College of Physicians (2018) guidelines for talking about dying do give the message that "fundamentally, we are dealing with people not pathologies" (p.5), and it is in light of this that consideration of treatment in the context of the illness experience should be key in decision-making (Bailey & Cogle, 2018). Galvin and Todres (2009) argue that human dimensions can be relegated in the list of clinical priorities, through an objective gaze, reducing the illness experience to measurable, quantifiable objects. Thus care is not sensitive to the holistic picture and the impact on the lifeworld that the LLC has had.

A doctor, whose elderly mother refused to have a life saving operation argued that:

> *"Death is no longer just in the hands of god or fate. It is often a decision. There is much discussion about the right of an individual to die, but not enough about the role of relatives and friends and the feelings of guilt, shame, ambivalence, regret, or uncertainty that may arise" (Black, 2018: p.1-2).*

The issues raised here have been brought into focus following a series of significant cases of disagreement between families and HCPs over the care of severely ill children, which have created headlines in the UK (Gallagher, 2017; Lagercrantz, 2018; The Lancet, 2017) The case of Charlie Guard (2016-2018) involved a protracted legal battle over the best course of action surrounding the continuation of treatment. The HCPs' verdict was upheld by the High Court but the parents chose to appeal this decision and found support from the public via social media campaigns and demonstrations, as well as from prominent global leaders such Pope Francis and President Trump. Following a further appeal, the initial decision was upheld by the Supreme Court and European Court. This case, along with the more recent Alfie Evans case (April 2018), highlight a degradation of trust between the public and HCPs in the making of decisions of this nature. As cases like these continue to capture public interest, there must be a sense of recognition from the healthcare profession that issues of trust and expertise within their own context are symptomatic of a broader societal trend where populism can be seen to hold sway over the public consciousness. It is within this narrative that the work of

HCPs with people with LLCs and their families needs to be appraised and assessed; failure to find a common language of experience could well result in a fundamental shift in the care relationship to the detriment of the profession as a whole. More must be done to ensure that there can be a more appropriate system of managing these complex decisions of care.

The difficult and stressful battle of fighting for treatment highlights the turbulent and painful journey for families when their child has a LLC. The emotional trauma and responsibility of fighting for your child's life on whatever scale, be it legal or conversational, portrays the intricate and deeply challenging task facing families and the HCPs. It should also be noted that these issues are not the preserve of paediatrics but remain prevalent across the lifespan for all that face death. Failing to effectively understand, on both the part of the HCP and the family, the concerns and priorities of the other, denies the opportunity for authentic holistic care that values personhood and lived experience. We run the risk of caricaturing and compartmentalising the positions of the respective parties in such a way as to negate the possibility of communication across the divide between the objective, medicalised gaze and the narrative of the individual. Perhaps attempts to bridge this divide can be understood as an imposition upon the remit of the other; consider the reaction of the HCP being given advice by a patient on how to do their job, or the parent of a dying child being told how to feel about the withdrawal of care. Again, without an attempt to establish humanised care, this division will continue to characterise our culture of care.

This breakdown in trust and communication in the case of Charlie Guard appears to be central, as well as the ethical dilemmas in making decisions about care. A children's cancer nurse leader on an international care ethics observatory panel with other experts reviewing the case of Charlie Guard suggested that clinical ethics teams should be available within a hospital for parents and carers who have to make difficult decisions with regards to withholding or withdrawing care (Gallagher, 2017). The nurse also believed that HCPs should have mandatory ongoing ethics and communication training to ensure that HCPs act within an ethical framework and to make sure that they communicate with families and carers in a sensitive manner (Gallagher, 2017). This is in order to avoid crisis points such as the case of Charlie Guard.

The interchangeable ways in which palliative care and end-of-life care are used by best practice governing bodies, HCPs, wider society, patients, and their families shows the confusion surrounding understanding 'care' when one is living with and dying with a LLC. This may be due to the way this care feels alien within the medical world in terms of preventing death; welcoming care that makes one think about their last few days of life and their dying experience is something which is fought against within the enlightened western world. A paucity in training for HCPs in palliative and end-of-life care shows the lack of emphasis that is placed on this period of time in one's life. From the review, training for HCPs needs to focus on communicating with people who have a LLC and providing assistance in judging when to have these conversations. This could be supported by a programme of regular supervision for HCPs, which could be assisted through the prioritising of continuation of care thus allowing for HCPs to build relationships with the people that they are caring for.

The proposed re-definition of palliative care can be seen to fail to confront the most pressing issue, namely the failure to reckon with the reality of the inevitability of death. In a world where science fights to conquer it, the acceptance of death needs to be recognised and incorporated culturally into our understanding of care, living in a way that acknowledges it and surrenders to its natural grasp. Addressing the stigmatised culture of this, in a movement towards accepting death, would make clearer the meanings of palliative care and the acceptance of discussing this when our time comes.

2.7 Conclusion

This literature review has synthesised and summarised research that has explored the meanings of living with a LLC as well as understanding the care provision available in the UK for this population. The illness experience of living with a LLC is complex and difficult for the person diagnosed and their families. With this said, there is a paucity of research that has approached understanding the illness experiences of a LLC from a subjective, idiographic perspective. In order to improve healthcare and for HCPs to get a more fulsome understanding of the holistic experience, the evidence base needs to place greater value on the nuances of living with an incurable illness. By understanding how illness impacts upon the lifeworld, those responsible can direct care to that which is more meaningful and sensitive to the sense-making of these experiences (Galvin, 2010). Although current care provision claims a holistic approach to care, it is evident from the research that HCPs feel uncertain and uncomfortable about communicating with people with a LLC and with the timing of when to talk about care that has connotations of death. This uncertainty can result in conversations being rushed and insensitive, leaving people feeling depersonalised. Care should aim to be more humanised and holistic in terms of seeking to understand the everyday experiences of what it is to live as a human being with an incurable illness. Understanding this from a lifeworld perspective can illuminate more about the vulnerabilities felt by people in this position and this can be used to direct care pathways, transcending professional categories and focusing on the subjective lived experiences of being human.

Chapter 3 – Methodological Approach

3.0 Introduction

This chapter will outline the methodological approaches used in this thesis to meet its aim of exploring the illness experience of living with NPC across the lifespan. This chapter begins by outlining the research paradigm used in this study and the ontological and epistemological positions that guided the decisions as to the methods chosen. It takes a reflective approach in telling the story of how this study changed from a pragmatic to an interpretivist paradigm as the programme of work evolved. A description of the strategies used in this thesis will be outlined, as well as providing a critical discussion of the methods chosen. Next, the analytical approaches will be addressed followed by an exploration of the techniques used to analyse the data gathered for the series of empirical studies conducted. The chapter will conclude with a discussion of the overall ethical considerations for this research.

3.1 The research paradigm

A research paradigm, or worldview, is a set of beliefs or assumptions held within research that explain how the world is made up (Guba & Lincoln, 1994). This consists of the ontological, which explores the nature of reality, and the epistemological, which concerns the nature of knowledge and directs and justifies the ways in which we gain knowledge (methodology) through the mode of inquiries we choose (Dures, Rumsey, Morris, & Gleeson, 2010; Guba & Lincoln, 1994). Generally, the two traditions to which researchers align themselves are positivism or interpretivism. The former is rooted in the scientific method, according to the Cartesian tradition, focusing on the generation of 'laws' based on causation, prediction and control (Blaikie, 2007; Smith, Harré, & van Langenhove, 1996). It is employed by researchers who wish to test claims and provide statements of them being either true or false (Ritchie & Lewis, 2003; Smith et al., 1996). This stance in research seeks to provide an objective view of the world, where there can be universalisation of laws. Thus the approach often favoured to test these claims and generate universal knowledge is rooted in hypothesis testing through experimental method. Such an approach seeks to isolate and manipulate variables in order to gather data that can be reduced to numbers for statistical analysis. These are often referred to as quantitative methods and are used to determine whether a change that is observed can be attributed to the manipulation of variables (van Langenhove, 1996). In spite of its popularity and utility within psychology and the social sciences, the positivist worldview lacks the tools to appreciate the wide range of human meanings, experiences and expectations that are brought to bear on a given event; the complex mix of individual and collective agency that characterises society (van Langenhove, 1996). Furthermore, it is impossible to explore meanings and complexities to life events when seeking to establish laws of behaviour, considering only a narrow view in the complex picture.

As one of the first to argue against this single-truth worldview, Immanuel Kant (1724-1804) suggested that reality cannot be reduced to a cause and effect explanation (Moran, 2000; Streubert & Carpenter, 2011). Developing this worldview, rooted in philosophy, interpretivism concerns itself with the understanding of human subjective experiences in the world. This approach seeks to understand human phenomena on a deeper level, embedded in human experience (MacKey, 2005). Through exploring patterns and meanings in contrast from set empirical markers, interpretative approaches seek to be open to experiential knowledge. This approach is convergent with qualitative research methods, those concerned with language and experience, in seeking to understand meanings in more detail. From an ontological perspective, when conducting qualitative research, researchers embrace the idea of multiple realities (Creswell, 2007). The commitment of most qualitative research is to get as close as possible to the participants being studied in order to explore *their* multiple realities.

A paradigm that views qualitative and quantitative methods as distinct but equitable ways of generating knowledge is pragmatism (Bishop, 2015). Many researchers have argued that there has been a successful convergence from an epistemological standpoint that has made combining qualitative and quantitative methods possible and popular when conducting health research (Bishop, 2015; Johnson & Onwuegbuzie, 2004; Morgan, 2007; Tashakkori & Teddlie, 2003; Feilzer, 2010). Johnson and Onwuegbuzie (2004) commented that pragmatism promotes “a needs-based or contingency approach to research method and concept selection” (p.17). This therefore removes the dichotomy of qualitative versus quantitative, alleviating this forced choice and allowing researchers to determine the most appropriate and effective approach in order to answer the research questions. Thus pragmatism provides a level of freedom in determining what might work without having to align with particular assumptions or without signing up to the sometimes closed mindsets of some of those working in qualitative or quantitative worlds. On the other hand, pragmatism fits with the progressive belief that the “consequences are more important than the process and therefore that ‘the ends justifies the means’” (Morgan, 2007: p.70). With regards to this study, this claim is contestable and will be discussed in more depth now.

3.1.2 A reflective consideration on the paradigm of this study

Initially, this study commenced with pragmatism as the central paradigm given that from the outset the project was likely to utilise a mixed methods approach and would therefore need to be able to accommodate the different epistemological positions that would be required. This necessitated a pragmatic and functional approach that would allow for the development and validation of disease-specific QoL scales for people living with NPC. This approach could therefore be seen as joining the increasingly popular mixed methods movement considered an emerging dominant paradigm in healthcare research (Bishop, 2015). In practice, this meant that the end goal of scale development was determining the theoretical and methodological decisions being made.

This had a particular impact given the substantial body of work that has been done developing a generic conceptualisation of QoL for use in such scales. My initial attempts at analysing the data I collected were predicated on an attempt to map out the data onto the traditional frameworks of understanding QoL that I had inherited from existing research. However, as the study progressed and as I spent more time in the 'field', attending the annual NPUK conferences and meeting people with NPC in their homes, I came to a greater understanding and appreciation of the participants and, as Creswell (2007) described, started to "know what they know" from first-hand information (Creswell & Tashakkori, 2007: p.18). The emerging picture began to challenge the legitimacy of using a more traditional method. Part of this understanding was to appreciate that this initial research trajectory was actually serving to provide barriers to my engagement with the participants, which was rooted in my preoccupation with the concept of QoL.

After careful reflection over time, I believed this was holding me back from getting to understand and explore the more ontological levels of meaning within the NPC illness experience. The traditional definitions of QoL were serving to cloud my understanding of, and potentially close my mind to, the specific phenomena I was studying, rather than bringing clarity. It therefore became necessary to jettison the inherited and generic conceptualisation of QoL and refocus my study purely on exploring, describing and making sense of the NPC illness experience in its own right. Freed from thinking about this in the confines of QoL removed distractions and enabled me to tune in to the accounts being conveyed. This also allowed for the recognition and consideration of contradictory descriptions of the experience of living with NPC without being tied to QoL. This therefore demanded a change in paradigm that would define the rest of the study. The freedom of the paradigm change was demonstrated in the nature of the research questions being posed as a result of my becoming further embedded within the NPC community. This led to an associated change in the direction of the thesis where an understanding of the lifespan became paramount. For example, this led to the gaining of ethical approval to interview people regarding their experience of a loved one dying to NPC. Thus, I would assert that the paradigm underlying this research changed through this study and was placed as interpretivist as opposed to pragmatic.

I still went on to meet with the requirements of my funders and to develop QoL scales for people living with NPC. However, as a study of lived experience, I decided not to operationalise the traditional understanding of QoL in terms of its definition or concepts used. Rather, stepping back from traditional QoL understandings and definitions allowed for a person-centred focus, thus living with NPC was conceptualised first and foremost as an ontological question of being. This novel approach to scale development will be discussed later on in this chapter and in chapter 7.

3.2 Theoretical underpinnings of this study

Having chosen the most appropriate paradigm and research questions, the next stage was to identify theories that might best help to explain the phenomenon of interest and the findings. Firstly, the ontological position of the current study aligns with the idiographic nature of phenomenology. Phenomenology as a theoretical framework provided an epistemological foundation informing the methodology of my study. A second theoretical underpinning used in this study was Galvin and Todres' (2013, 2011) existential-phenomenological theory of the lifeworld, which sought to explore further the ways in which living with illness impacted upon a person's lifeworld. Thus, this thesis conceptualised living with NPC as an ontological question of being and concerned itself with the meanings attributed to this experience across the lifespan. In keeping with the epistemological positioning of this study, methods related to phenomenology were used, focusing on the lived experiences and sense-making of individuals. Thus, this reflects a coherence between the levels of decisions made in with regards to the thesis' paradigm, methodology and method (Yardley, 2000).

3.2.1 Phenomenology

Historically, phenomenology as a philosophy presented a challenge to the scientific method which has in turn evolved as an alternative traditionally used within the social sciences. The separation from the natural sciences and the move towards differing ontological and epistemological approaches was emphasised by this "*radical* way of doing philosophy" through phenomenology (Moran, 2000: p.4; Pascal, 2010). Moran (2000) writes:

> *"Phenomenology is best understood as a radical, anti-traditional style of philosophising, which emphasises the attempt to get to the truth of matters, to describe phenomena, in the broadest sense as whatever appears in the manner in which it appears, that is as it manifests itself to consciousness, to the experiencer." (p.4).*

Many agree that Edmund Husserl (1859-1938) was the founding father and transformer of phenomenology, reconsidering the nature of psychology as a *science* by bringing in a philosophical foundation (Moran, 2000). Following Brentano (1838-1917), Husserl's phenomenology concerned itself with the conscious lived experience of phenomena as perceived in daily life (Crotty, 1998; Moustakas, 1994). Husserl suggested that in order to understand human experience we must explore the lifeworld (*lebenswelt*) of an individual, the beginning and end point of existence (Galvin & Todres, 2013). Husserl believed that it is important to set aside fore-assumptions and theories and get 'back to the things themselves' (*sachen selbst*) as they seem to us in the everyday (Moran, 2000). In relation to this, a central component to Husserl's philosophy is the notion of intentionality, the internal experience of consciousness *of* something, an object that is personal in a relational and meaningful context (Moustakas, 1994; Wertz, 2005). This notion goes against the Cartesian tradition of dualism, in that it suggests that the mind-body and subject-object coincide. There is something inherent in our experience that is intertwined with the world and how we perceive it; we are not separate from the world and our perception of it carries meaning.

Husserl's beliefs require us to take a 'phenomenological attitude' (Finlay, 2009), which suggests steps should be taken to identify and isolate the phenomenon in question. One of the ways to do this was by practising *Epoché* through bracketing. Through bracketing the researcher's own subjective understandings; prior knowledge such as "scientific, philosophical cultural and everyday assumptions" should be put aside, which can then provide a fuller understanding of the phenomena in question (Moran, 2000: p.11).

Martin Heidegger (1889-1976), a student of Husserl, focused more ontologically as opposed to Husserl's firmly epistemological approaches (Reiners, 2012). Heidegger suggested that the principal of phenomenology was the meaning of *Being* and that this could not be separated from human beings existing together in the world (Moran, 2000; Van Manen, 1990). Heidegger disagreed with Husserl's focus on intentionality and argued that it is impossible to bracket our presuppositions. Doing this would "shed our experience" and in turn we would lose our capacity to provide a context for understanding others' experiences (Pascal, 2010: p.3). For Heidegger, we cannot prevent our own perspective from colouring experiences, and our interpretations of the experience of others. Agreeing with Husserl in that we must focus on the everyday or ordinary nature of experience, Heidegger rejected the view that detailed description was enough on its own and instead thought that it was necessary for one to move past the description of phenomena and rather sought to understand the meanings that are embedded in everyday occurrences.

Heidegger described how the way in which we relate to the world functions within a 'hermeneutic circle' of interpretation based on knowledge from our previous experiences, placing value on these and ourselves as *being-in-the-world* (Pascal, 2010). Heidegger did not develop a method for phenomenological research but 'Heideggerian' methods have been developed based on a hermeneutical approach (Schmidt, 2006). This concerns the means of achieving an understanding of human experience and how we see meaning in the world around us through the interpretation of our experiences. It encompasses the combination of experiences portrayed by a participant along with the experiences of the researcher (Koch, 1996). Crotty (1998) importantly acknowledged that unless there is a level of accountability in the use of hermeneutics as a method, it is open to critique on the rigour and validity of the research conducted. This involves an openness on the part of the researcher to be transparent regarding the influences that have stoked the interpretation (Dahlberg, Dahlberg, & Nyström, 2008).

Utilising hermeneutics in research helps to understand and present the non-static nature of our existence in the world and the various levels of meanings that are found (Conroy, 2003); particularly in this context, the meanings attributed to living with an incurable illness. This process also served to make explicit the meaning-making process, thus building a further level of rigour. Conroy (2003) suggested that hermeneutics transfers us from a stationary viewpoint to one of perpetual flux. This viewpoint is one that is intertwined with the lives of others, where our knowledge and understanding of the world is not fashioned by an individualised construction but instead is founded on a build-up of the merging of interpretations. Svenaeus (2000a) suggested that:

> *"…theories of phenomenology and hermeneutics are particularly suitable for this kind of analysis in the case of health and medicine, since they manage to explicate features that would otherwise remain hidden in an exclusively natural scientific approach to the subjects." (p.6).*

The use of interpretative phenomenology in this thesis enabled a more detailed understanding of the illness experience, which has not been understood before in people living with NPC in the UK. Taking an interpretative Heideggerian hermeneutical approach enabled me to understand the interrelations of living with illness in a more meaningful way, which is based on gaining understanding of an individual's perceptions of these realities. In addition to the intersubjective experiences of being-in-the-world, Merleau-Ponty (1908-1961), a successor to Husserl was keen to emphasise the situated nature of human experience but with a distinctive focus on our embodiment and the role of the body in interpretation. Merleau-Ponty expressed the intimate relation of body and world. He said that humans always interpret others through knowledge of their bodies and used the term ‘body-subject’ to refer to humans as interpreters in this way: “Our own body (*le corps propre*) is in the world as the heart is in the organism; it keeps the visible spectacle constantly alive, it breathes life into it and sustains it inwardly, and with it forms a system” (Finlay, 2006) (Merleau-Ponty, 1945/1962: 235). Further to this, Meleau-Ponty recognises the relationship between chronological and historical situatedness and the individual’s being-in the world. This, in turn, serves to add a further dimension to our conception of the lifeworld as we seek to explain the lived experiences of how we understand and experience the world (Mishler, 1984).

Therefore, attempting to understand the lifeworld can be seen as an important approach to gaining a clearer glimpse of the world that we experience and its many levels of meaning. Mishler (1984) explained that the voice of the lifeworld is characterised by “…contextually-grounded experiences of events and problems... These are reports and descriptions of the world of everyday life expressed from the perspective of a ‘natural attitude’” (p.104). The work of Galvin and Todres (2013, 2011) suggested a theory based on the philosophical foundations of Husserl, Heidegger and Merleau-Ponty, providing an understanding of human experience that will underpin this thesis alongside phenomenology.

3.2.2 Lifeworld theory

As described in chapter 1, the lifeworld encompasses our relationship with the world and our meaning-making (Mishler, 1984). Accepting the notion that the body is a fundamental component of a person’s lifeworld, one can assume that the lifeworld changes when the body changes. This is particularly relevant when one becomes ill, or in the case of NPC, when the illness is incurable. Phenomenologists have attempted to thematise interrelated dimensions of the lifeworld in order to better explain it (Ashworth, 2006, 2003). Lifeworld-led theory, which has been developed by a variety of different proposals and interpretations (Ashworth, 2006, 2003) has been used to explore people living with illnesses (Ashworth, 2006; Smith & Shaw, 2016; Todres & Galvin, 2006) as well as its utility and operationalisation in understanding healthcare (Galvin et al., 2018).

This study will be underpinned by Galvin and Todres’ (2013, 2011) lifeworld theory due to their exposition of the lifeworld complementing the phenomenological interpretivist research paradigm that this thesis is built on. Galvin and Todres’ (2013, 2011) theory consists of six dimensions, which help to generate an understanding of the lifeworld, capturing a fuller description of human experience. These are: temporality, spatiality, intersubjectivity, embodiment, identity and mood. Galvin and Todres (2011) suggested that these domains are intertwined with one another, yet hold certain nuances of lived experiences.

Temporality

The notion of temporality can be understood as the human conception and experience of time. This is particularly related to the narrative framing of one's experience and one's understanding of the past, present and future. Time has the capacity to open up or close in on and individual's sense of self and purpose; it provides the rhythms and routines of the lived experience. It is a framework through which we understand the world around us. When in the context of illness, the past may be desired in the time before disease was diagnosed and the future may feel empty of opportunity. This has significant ramifications for how an individual views their life when they have a life limiting illness and the extent to which these differ to how a carer thinks about the illness in relation to time.

Spatiality

Spatiality is best understood as the relationship with the lived environment. The meaning significance and feelings associated with place and space can drive and evoke a range of interpretations that matter to the human experience. These experiences are inevitably influenced by the associations individuals make with a particular environment; feelings of distance, closeness, assurance or enclosure are common experiential components of such reflections. Environmental changes to one's home if their mobility deteriorates may evoke feelings of disorientations and confusion. A person moving into respite care or a hospice may experience a degree of disconnect from that which is meaningful to them. For example, a child living with a life limiting illness may need to move from having their bedroom upstairs due to them not being able to climb the stairs anymore; leading to disorientation and confusion.

Intersubjectivity

Intersubjectivity refers to how we are in a world with others; our lives take place within a social world. We can share experiences and meanings with others and relationally, others can relate expressed meanings of their experiences; "through intersubjectivity and language, we locate ourselves meaningfully in the ongoing interpersonal world" (Todres, Galvin, & Dahlberg, 2007: p.57). In order to understand in a fuller way the illness experience, the meanings placed on relationships and culture is vital. Understanding a person's interpersonal world in the context of their illness experience may be central to understanding and illuminating the lifeworld of the individual, as diagnosis with such an illness has an impact upon relationships (Strauss, 1975, cited in Bury, 1982).

Embodiment

Embodiment refers to the lived relationship between our concrete physical bodies and the world around us; our bodies as conduits for lived experience. Here the notion of 'feeling' in such a way that transcends mere biological reactions to stimuli becomes an important feature to understand. Our physical, bodily reactions to the world are significant; our bodies become the lens through which our interaction with that which is around us is perceived. In the context of illness, the embodied framing and response to limitation, finitude and symbolism become vital components of lived experience. Such a sense of embodiment helps us to understand illness in a more complete way through our bodily interactions within the world.

Identity

Identity refers to feeling at one with the world. It encompasses our interpersonal relationships and our interactions with society and how they affect our self-growth. In the context of illness, it is an incredibly important element to focus on in terms of how illness impacts upon an individual's identity; how one's identity forms or changes when living with a life limiting illness and the disruption illness can bring to our identity (Charmaz, 1995). Yet, it is important not to assume that changes to identity during illness are always unwelcomed. Identity is also an important component part of our social existence, that which provides a framework for our interaction with others (Charmaz, 1995).

Mood

Mood is an element that influences each of the other dimensions of the lifeworld. It creates a perspective from which one views one's spatial, temporal, intersubjective and embodied experience can be understood. Mood often goes beyond the intricacies of human experience, language and interaction and characterises an experience as a whole. It has the capacity to determine our priorities and values, to motivate or demotivate and to determine our sense of direction. It can therefore be seen as an important holistic component of understanding both health and illness; a subjective platform from which experience is built. Illness can cause a sense of 'unhomelikeness' (Svenaeus, 2000c) from how one may feel in one's body relationally with others within the time and space they are in.

Translated into a methodology, phenomenology in combination with lifeworld theory utilises Heideggerian notions of being-in-the-world with others in the space and time that we live in. This exploration is holistic in nature, making sense of the 'whole' in lived-experiences of those who are impacted by NPC. Living with NPC is an existential matter in that it is an incurable illness so it therefore raises issues surrounding life and death thus calling for a reflection on the nature of being (Heidegger, 1962/2000). Thus, this theory offers an appropriate mode through which to investigate the phenomenon of living with NPC in a way that embraces the depth and richness required to fully understand such a critical existential issue. The next section will outline the phenomena of what this research is exploring as well as the strategies that will be used to explore this.

3.3 Phenomenon of the study

The theoretical underpinnings discussed above framed the inquiry within the deep-rooted meanings of human phenomena. As a result, this thesis sought a multiperspectival outlook (Larkin, Shaw, & Flowers, 2018) to understand the experiences of living with an incurable illness across the lifespan, exploring children's and adults' experiences of living with NPC as well as their family caregivers. In addition to this, an exploration was conducted into the final period of dying with NPC.

The main aim was to look at the sense-making of people diagnosed with NPC first and foremost. If this was not possible then the sense-making of the family caregivers as a secondary and supplementary perspective. For the studies focusing on child and adult experiences of living with NPC, if a person diagnosed with NPC was not able to take part in the interview due to either being too young or due to the cognitive effects that NPC had on their communicative skills, then family carers were asked to describe the experiences from the perspective of the person they were caring for. They would also discuss the experience from their perspective, which presented a fuller description of what it means to live with NPC as a family. Additionally, in the study that explored the phenomenon of dying with NPC, bereaved parents discussed their experiences of end of life with NPC. Families conveyed the narrative from their perspective, but would also communicate how they believe their dying child experienced these moments. Therefore, it should be recognised that when necessary, this sense-making can be understood through the simultaneous consideration of both first and second hand accounts. The first-hand account is the experience of the person diagnosed with NPC, whilst the second-hand account is the parents/carers conveying the experience of the people with NPC that they are caring for. This can be defined as a double, and sometime triple, hermeneutical approach. This is supplemented by parents/carers also providing an account of their own experience of being impacted by NPC, which would be their first-hand account. This therefore demonstrates that there is a consistent hermeneutical approach that runs through each of the studies. Included in this notion is the conviction that a multiperspectival vision of the illness experience is both desirable and achievable through the careful and considered reflection of both caregivers and researcher, even when the capacity of the participants is limited.

This is supported by Larkin et al. (2018) who argued that adopting a triangular understanding of different viewpoints can be a more powerful analysis than one that is drawn from a single sample, using the multiplicity of people's stories. This approach is especially pertinent for this study due to the closeness in relationships between persons living with NPC and their reliance on their family members to provide care and support. This leads to a greater understanding of the lived experiences of the family's "multiverse" (Larkin et al., 2018: p.13). In addition to this, including people with NPC in this study provided a way of gaining a level of understanding of their experience of living with illness. The neurological manifestations of NPC may sometimes impede upon a clear understanding of the meaning of what is being communicated. Having their family carers present may provide a level of support in helping them to communicate what they would like to. However, as explained below, strategies and analytical techniques will be used which steer the study in such a way as to grasp more fully the experiences of living with NPC.

3.4 Strategies of inquiry

Epistemologically, I assumed an approach that at first glance might seem contradictory in that it was both inductive and theory-driven because of its commitment to describing and understanding the lifeworld in its wholeness. Working from an interpretivist phenomenological paradigm illuminated the deeper-rooted, internal meanings of the illness experience and provided a contextual and informed understanding of the phenomenon of living with NPC (Merleau-Ponty, 1962). The strategies chosen to guide the epistemological axis of this study align closely with its theoretical underpinnings. These strategies are: reflexivity and an abductive reasoned logic of inquiry. These strategies will be fundamental in how this study is conducted.

3.4.1 Reflexivity

Throughout this study, I embedded reflexivity into the research process for several reasons. Firstly, reflexivity provides a strategy for enabling transparency in how I as the researcher conducted the programme of work including the generation of data and its analysis, showing that preconceived understandings are identified and considered (Shaw, 2010). As we are living beings, the way in which we view the world will be shaped by the culture in which we have been raised. The cultural context of all parties involved in the process will inevitably influence the relationships that are built and the way they are portrayed in a research setting. This means that the lived experiences that I carry with me must be understood to be in dialogue with the experiences of all those engaged in the research (Shaw, 2010). Through taking part in reflexivity, we are "making ourselves aware of our own feelings about and expectations of the research…" (Shaw, 2010: p.235). By doing this, we are acting vigilantly in identifying any fore-understandings and ensuring that these are carefully considered throughout the research inquiry. The practice of reflexivity interweaves with the Heideggerian theoretical underpinning of this study in *dasein* - our very being - meaningfully connected to our context (Goldspink & Engward, 2018) thus strengthening the research by acknowledging and discussing these connections in a meaningful way.

The practice of reflexivity meets with the hermeneutic commitment of this study, in the back-and-forth pattern between the language used in interacting with participants and the impact this had upon my own conceptualisations and experiences, awakening "intellectual vistas" from which to ascertain and explore subjective understandings (Goldspink & Engward, 2018: p.12). Thus, reflexivity enabled me to have a method of ensuring that I was careful in how I approached interviewing participants and doing analysis, being self-aware of my own personal experiences. Goldspink and Engward (2018) commented:

> *"Through reading, thinking, listening, and notably writing, reflexivity emerges as an active conversation between the raw data and the researcher's past and present self, unwrapping questions to recognise and then progress 'taken-for-granted' appraisals." (p.12).*

As Shaw (2010) explained "By engaging in reflexivity, that is, proactively exploring our *self* at the start of our research inquiry, we can enter into a dialogue with participants and use each participant's presentation of self to help revise our fore-understanding and come to make sense of the phenomenon anew." (p.238). The bond between phenomenological investigation and my own engagement with writing makes visible the connections between my expression of thoughts and the participant's voice (Van Manen, 2006). Thus, the importance of documenting my thoughts throughout the study's entirety was a meaningful component of the research itself and is why this is classed as a strategy in conducting the series of studies presented.

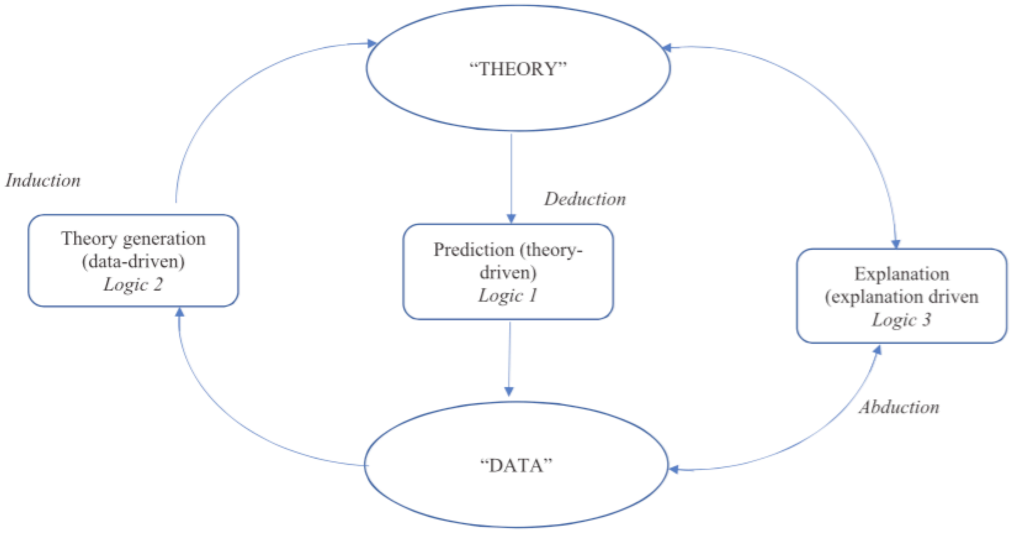
With that said, and in order to contextualise how I embarked upon this research, I had both personal experiences of caring for a loved one with a LLC, through my father's experience of Alzheimer's disease, and professional experience in my previous research work, which had involved other neurodegenerative diseases. Through my experience of caring for my father for 11 years, I entered the study with relevant personal experience, as well as my own personal accounts of the interruptions illness had caused to my own and my family's lifeworld. Reflexivity was crucial for me in finding the balance between allowing my own experiences to come into play but not overshadowing the experiences of others. Reflecting on this, I arguably bracketed too much in my early interactions, not in the sense of totally discounting my own experiences, but rather in being so careful to interpret people's experiences from what they said, that it served to silence the resonance between our lifeworlds. However, as I grew in confidence with my research, as I think is highlighted in the progression of my empirical studies, in my last study I was able to utilise embodied interpretation, which welcomes these resonances. This accomplished a "working in the light of the data to overcome the shadows present" in my own fore-understandings (Goldspink & Engward, 2018: p.12) and this will be discussed further in the methods section below when presenting the interview techniques.

3.4.2 Logics of inquiry

Hiles (2012) argued that the term 'mixed methods' should be replaced with the term "mixed designs" as he suggested that methods is restrictive in that it refers to purely collecting data, thus losing the importance of examining the underlying paradigmatic assumptions, which in turn formulate the research questions and the understanding of the logic of inquiry (Hiles, 2012: p.7). To move away from this, he argued that the terms qualitative, quantitative and mixed methods should be replaced with data-, theory- and explanation-driven logics of inquiry respectively. This is because the terms qualitative and quantitative characterise opposing traditions of inquiry, but more than this, Hiles (2012) argued that the disparities transcend the type of data one is collecting and instead refers, at a more fundamental level, to the types of inquiry listed above. Epistemology should drive the question forward, not the narrow confines of method. Hiles (2012) deciphers the difference between the logics of inquiry. Both theory-driven and explanation-driven inquiry are *a priori* but explanation-driven inquiry is different in that it does not deduce to make predictions. Explanation-driven inquiry is also exploratory in the same way as data-driven inquiry, but the theory does not emerge from the data, it exists *a priori.* As Hiles (2012) explained, the explanation-driven logic of inquiry is about finding the "best explanation" of the data (p.6). The relationships between theory and data is illustrated in Figure 2.

The emphasis of this thesis was to understand and explain the experiences of living with NPC. This drove the logic of inquiry to be explanation-driven in that it was led by the inductive analysis of the data, and the lifeworld theory was used subsequently to help find the best explanation of the findings. Within the worldview of interpretavism, the emphasis is on the inductive, data-driven understanding of experiences, therefore questions arise as to whether the adoption of theories within this paradigm broadens or limits our understanding of the data. My rationale for including Galvin and Todres' (2013, 2011) lifeworld theory in this study is that it is founded on a phenomenological understanding of the lifeworld and is fluid in its outlook. I would argue that including this theory widened the understanding of living with illness across the lifespan. Using an existential framework was an appropriate way of helping to better explain the findings.

Figure 2. The relation between theory, data and logic of inquiry in Shaw, Hiles, West, Holland and Gwyther (2018).



This explanation-driven logic of inquiry works well when studying a topic/concept/experience in considerable depth and theories are used to illuminate the understanding of the research experience as seen in this study (Hiles, 2012). Kvigne, Gjengedal and Kirkevold (2002) argued that the use of theoretical concepts enriches understanding in a way that acts a springboard to knowledge. Thus theories can assist in leading the researcher to more-enlightened perceptions behind the primary descriptions of data. This may of course have the reverse effect, where one may disqualify the theory based on the data gathered. Yet the utilisation of the theory in the first place is likely to improve the researcher's exploration. In tune with the previous section, I embarked upon an epistemological reflexive endeavour and all of these issues were discussed in-depth with my supervisors. Exploring these issues was an active element of the analysis in conducting the programme of work.

Therefore this explanation-driven logic of inquiry encompasses examining ideas principally from an inductive perspective but in hand with abductive logic, thus bringing together data and theory to broaden the scope of our understanding of living with NPC and its impact on the lifeworld. This process complements the phenomenological hermeneutic circle of interpretation (Hoy, 1999) and involves a continuous movement and open dialogue between data, theory and experience thus the strategies here (reflexivity and abductive reasoning) fit hand in hand in its dialogic arc (Pascal, 2010; Ricoeur, 1995). These strategies provided an epistemological foundation for informing the methodology. Such inquiry sought to prioritise the holistic experiences of illness - being-with-illness - in keeping with the ontological position of this thesis.

3.5 Methods

This section will discuss the practicalities of this research in terms of data collection, analytical approaches and analytical techniques.

Interviews attempt to understand the world from the participants' points of view in order to reveal the meanings present in their lived world (Kvale, 2007). I chose in-depth, biographical interviews as a data collection method for their methodological accord with the interpretative stance of this thesis in understanding the experiences of living with and dying from NPC. The interview acts as an instrument for opening and understanding the situation that people are experiencing, which in the context of this study was life with incurable illness (Kvale, 2007). The biographical interview technique was viewed as the most appropriate method of providing a more natural narrative of the lifeworld that is led by the participant as opposed to the researcher. This enabled a platform for people to share their experiences in a way that may help people to feel liberated.

Van Manen (1997) contended that the fundamental research questions for initiating the inquiry should direct the choice of the type of interview that is most suitable. As explained above the initial aim for meeting with the funder's brief was to develop a disease-specific QoL scale for people living with NPC, meaning the ethical agreement had already been established prior to me starting my PhD. This meant that the interview schedule had already been developed. Yet, in the same way that my paradigmatic foundation shifted as I developed my understanding of and confidence in the phenomenon, so my methodological approach adapted and evolved to better reveal the data I sought. The interview schedule and the prompts were therefore used less and less as I wanted to focus on the experiences of living with NPC and not be in the shadow of QoL. Further to this, as I grew in confidence the interviews went from a semi-structured format to a biographical one as the scope of the study changed to focusing on people telling their stories of living with NPC, using only a couple of prompts from the original interview schedule. This occurred early on in the data collection process as it became increasingly noticeable that participants needed the space to share their experiences without much prompt. The freedom of the biographical interview offered the participants the opportunity to talk at length and give greater detail about their experiences of living with NPC. This shift in focus to understanding the illness experiences was highlighted when I applied for ethical agreement to conduct a study looking at end-of-life care and the dying experience of someone with NPC (chapter 6).

The over-arching interview question for the first two empirical studies focusing on the children (chapter 4) and adults (chapter 5) were: "What is your experience of living with NPC?" with further exploration of issues such as the diagnosis journey, daily life and changes over time. The last empirical study focused on the dying experience of a child with NPC, with exploration into the diagnosis story, thoughts surrounding palliative care, the dying process and life after death. In general, participants would naturally take a chronological explanatory order starting from the beginning, when symptoms appeared, to the diagnosis period and moving into how life had changed and their subsequent experience. The argument for choosing a biographical approach was that participants would speak about what was important to them within the experiences of living with NPC, therefore encouraging discussion of the issues and events that had the greatest impact upon their lifeworld.

Van Manen (1997) suggested that the job of the researcher in interviews that are steered by hermeneutics as its worldview, is to maintain the participant's focus on the matters being explored. Although the interviewees had 'free reign' to talk about their experiences and their sense-making of living and dying with an incurable disease, the interviewer remained attentive to this, giving meaningful responses to their experiences. More structure was offered if it was needed. For example, some of the adults with NPC who were interviewed valued slightly more structure whereby I would ask a few questions that would broadly follow a chronological path of events as mentioned above. Yet this still retained flexibility in how the responses unfolded as participants led the direction of the conversation. The questions that were asked were very open-ended, thus the participant could navigate them in the ways that were most meaningful to them. When interviewing family carers, I sometimes reminded them within the interview to put themselves in the shoes of the people they were looking after and how they made sense of how the person with NPC made sense of their experience.

Conducting interviews required a considered approach in acting with sensitivity for the people who were being interviewed. A reflection prior to each interview allowed me to be thorough and mindful of the scope of variability between people participating. The interviewer showing a level of understanding can enhance the interview and thus enrich the data. This can be achieved by establishing good dialogue, which is the responsibility of the researcher and can enable a rapport by helping the participant feel at-ease (Dahlberg et al., 2008; Kvale, 1996). Kvigne et al. (2002) suggest that contributing factors such as the tone and body language influence the depth that participants will go into, thus influencing the richness of the data being collected. These factors will distinguish "whether the informant opens her lifeworld for the researcher or not" (p.65) arguing that this necessitates the researcher to have "not only an open mind, but an open heart" (Kvigne et al., 2002: p.65).

Reflexive practice was key here in working out how best to approach the interview in terms of the dialogue and exchange that would take place between researcher and participants in understanding their experiences. My presuppositions were based on my experience of being a carer and also on the responses of others towards my illness experience. I found that in many cases, people would try to cover silences, a behaviour which may be found in interviewers, even subconsciously, especially when the topic being discussed is sensitive and difficult to hear. Reflection on this helped me to pause during the interview process, whilst the participant thought about what they wanted to talk about next. I chose not to outwardly exchange much from my own story early on in the course of conducting interviews as it became evident that people living with rare disease feel undervalued and shifting the focus onto me may only have served to exacerbate this problem. I wanted to give participants the platform to share *their* experiences. On some occasions I asked if certain instances had made the participant feel a particular way, inadvertently using my own experiences to guide and frame the conversation. Following the interview, I would document the moments of shared experiences that occurred where I felt a degree of familiarity with aspects of participants' stories. This was not limited to the times where I felt the chiming of familiarity, as useful reflection was also gleaned in the moments of disconnection, which were equally useful to the development of understanding. After each interview, I spent time reflecting on how the interview had gone and wrote a short summary of my own thoughts and feelings of what struck me as pertinent, my reactions to what participants had said and whether I would do anything differently in my next interview. These moments would be reflected on especially in the analysis, which will be spoken about next.

3.6 Analytical approaches

This study had two analytical approaches to understand the experiences of living with and dying from NPC. Firstly, a hermeneutic-phenomenological analytical approach was adopted, which analysed the data collected inductively. Secondly, the lifeworld theory was used to explore data further. These approaches drove the analysis in keeping with the phenomenological paradigm of this study in that they both embrace hermeneutics, involving a continuous movement between theory, data and experience.

3.6.1 Hermeneutic principles

As elicited previously in this chapter, the hermeneutic approach is usually performed using the metaphor of the hermeneutic circle, to describe the open-dialogue between the parts and the whole (Koch, 1996). In order to understand the phenomenon in question, the researcher's prerogative is the repeated process of moving between interpretations of the text in the sense that understanding the whole requires grasping its parts and "comprehending the meaning of the parts dividing the whole" (Crotty, 1998: p.92).

Conroy (2003) derived from her own research work and a critical examination of philosophical literature, a list of hermeneutical principles that should be considered in research. A number of these principles were followed in this study in order to understand the experiences of people impacted by NPC. Conroy (2003) encouraged the researcher to access the participants' meaningful world, by immersing ourselves into it, both when present with the participant and when analysing the data (Benner, 1994). The union between hermeneutics and reflexivity is revealed in the researcher making transparent the shared world of understanding. Thus, the researcher sheds light on their 'background' understandings and why the researcher may be drawing out particular elements of the participant's narrative account. The techniques used to do this will be discussed later on in this chapter but this practice is reliant on maintaining a repetitive questioning approach in the quest for uncovering meaning-making in the participant's experiences (Conroy, 2003).

3.6.2 Dwelling-mobility lattices

Along with their lifeworld theory, Galvin and Todres (2013) created two, interrelating existential descriptions of well-being and suffering which draw on the elements in their lifeworld theory. These descriptions are founded from Heidegger's proposal of humans feeling an at-homeness with what is, whilst also being oriented towards future possibilities, bringing together Heidegger's notion of *'Gegnet'* in dwelling and mobility respectively. In this view of well-being, the idea of 'dwelling' is grounded in the present and a peaceful acceptance of this position, a sense of 'coming home' to what we have been given; the essence of 'mobility' is the feeling of future possibilities, with one's body and with others (Todres & Galvin, 2006). This combines the peace of dwelling with a feeling of freedom of movement towards the future, encompassing "a feeling of rootedness and flow, peace and possibility" (Galvin & Todres, 2011: p.65), *Gegnet*. In relation to existential suffering, dwelling represents homelessness and unsettledness. In the essence of mobility, suffering represents a permanent feeling of imprisonment and entrapment. Finding a sense of *Gegnet* can be explored through understanding the lifeworld of the person, their lived experience and their journey towards being-at-home. This is pertinent when looking at the context of changes to the lifeworld of a person living with an incurable illness, where in some ways one feels at-home in the body that they have always known, but also, homeless, as the body changes and becomes dis-eased (Toombs, 1993). Galvin and Todres (2013) suggest well-being and suffering are a continuum and are in relation to one another, and as embodied experiences hold multiple qualities simultaneously. They argue that a dual understanding of both well-being and suffering within a care setting can meet individuals in their "vulnerabilities" and their "possible freedom" (Galvin & Todres, 2013: p.98). These different levels of well-being and suffering are shown in the lattices in table 3.1 and 3.2. The different levels of well-being and suffering do not have to be experienced in their fullest sense to be afforded to human existence; there can be different levels of both, dependent upon the focus and emphasis (Galvin & Todres, 2013). These lattices were used to derive meaning from this co-existence of suffering and well-being whilst being in dialogue with inductive interpretations from biographical interviews, in order to further examine the meanings of living with NPC.

3.7 Analytical techniques

This study used a pluralistic approach to data analysis. This section will outline two analytical techniques that were used in this programme of work to analyse the data inductively. A rationale for their use is provided.

3.7.1 Interpretative Phenomenological Analysis

In keeping with the phenomenological worldview of this study, Interpretative Phenomenological Analysis (IPA; Smith, Flowers, & Larkin, 2009) was used as an analytical technique. The theoretical orientation of IPA fits within the paradigm because it is consistent with phenomenological principles in seeking to make sense of the meanings individuals attach to being human and their experiences (Smith et al., 2009). IPA was developed from Heidegger's work, which draws upon interest influenced by the interpretations of language, objects, relationships and time (Smith et al., 2009). IPA is strongly influenced by hermeneutics in its recognition of the importance of interpretation; this provides an insider's perspective, which is central to this study's objectives (Brocki & Wearden, 2006) and in keeping with the hermeneutical strategy that this study is encompassed within.

Table 1. 'Dwelling-mobility lattice' adapted from Galvin and Todres (2011) theory of well-being.

Element of the lifeworld	Mobility	Dwelling	Dwelling-mobility
Spatiality	**Adventurous horizons** Where spatial opportunities bring a sense of adventure. Possibilities that offer movement (metaphorically or literally)	**At homeness** A sense of being at-home and feeling of settledness within the familiar surroundings	**Abiding expanse** Both feelings of at-homeness with opportunities for adventure, expanding horizons.
Temporality	**Future orientation** Being excited about the future, what is a head. Not being trapped or stuck	**Present-centredness** Settled in the present in a desirable way. A sense of belonging is felt.	**Renewal** Fusion of future possibilities with contentment for the present. Being satisfied with the now and a welcome readiness for the future
Inter-subjectivity	**Mysterious inter-personal attraction** In tune with interactional possibilities. Understanding the mystery of others	**Kinship and belonging** An natural being together; 'we' rather than 'I' and 'you'	**Mutual complementarity** Both a togetherness and excitement at learning and embracing the new
Mood	**Excitement or desire** Sense of attunement and optimism of movement.	**Peacefulness** Stillness where there is a welcomed silence and acceptance of things, a 'letting be'.	**Mirror-like multi-dimensional fullness** Mood which captures fullness and energy of moving forwards but also with a at-homeness within the world and sense of self
Identity	**I can** Being able to. Experiencing oneself as being on the move (literally or metaphorically)	**I am** A self that it supported by histories and contexts that fit with who 'I am'	**Layered continuity** A continuous sense of 'I can' and a strong sense of 'just being'. Creating a sense of ontological security
Embodiment	**Vitality** An embodied energy that offers freedom to move and possibility	**Comfort** Feelings of comfort and relaxation. Sense of familiarity and closeness with one's body.	**Grounded vibrancy** An energised movement and a sense of feeling at-home within one's body. Both 'being' and 'becoming' is possible

Table 2. ‘Dwelling-mobility lattice’ adapted from Galvin and Todres (2011) theory suffering

Element of the lifeworld	Mobility	Dwelling	Dwelling-Mobility
Spatiality	***Imprisoned*** Feelings of hemmed in, unable to move, trapped with no room or any horizon that can give respite	***Exiled*** Cast out or wrenched from a familiar place. A sense of banishment with limited opportunities	***Roomless*** One longs for home but is trapped in homelessness – there is little or no possibility
Temporality	***Blocked future*** Stuck, or cut off from any sense of future. Feelings of frozen in time	***Elusive present*** Temporal unsettledness – life passing oneself by. A sense of looking backwards to ‘ghosts and fears’	***No respite*** One is lost in limbo constituting of anguish and despair. There is no way forward
Intersubjectivity	***Aversion*** An aversion of being with another or others. A sense of interpersonal conflict	***Alienated isolation*** One’s sense of interpersonal belonging and kinship is ruptured. Feelings of cast out and exiled from interpersonal warmth.	***Persecution*** Feelings of threat from others with no way out. An interpersonal trust being lost.
Mood	***Depression*** ‘Closed in’ mood of limited or dark horizons. Lack of energy and motivation, a pessimistic outlook a sense of not being able to carry on.	***Agitation*** Emphasises the mood of unsettled restlessness by a feeling of irritation, anxiety and disturbance. Feelings of homelessness and a rupture to dwelling.	***Restless gloom*** One may feel a gloom that is intolerable where one must escape, characterised by a desire to end everything.
Identity	***I am unable*** ‘I lack ability.’ Feelings of uselessness and failure – a sense of ‘not being able to.’	***I am an object or ‘thing’*** Being identified by self or others as ‘a thing’ or an object. Feelings of being turned into something or someone else – its essence being injured.	***I am fragmented*** One’s sense of self is overcome, even possessed by forces that feel alien or ‘other.’
Embodiment	***Stasis and exhaustion*** A physical suffering that is characterised by the body’s inability or lack of desire to move, or a felt sense of impaired or threatened bodily functions.	***Bodily discomfort and pain*** A physical suffering that is characterised by bodily discomfort and pain. Here it is difficult to dwell or feel at home in one’s body because of a palpable sensation that something is wrong.	***Painful closing down*** *There is a whole spectrum of suffering. Bodily suffering may feel unspeakable in that there are few shared words that can indicate the convergence of both extreme ‘discomfort/pain’ and extreme ‘closing down’ of bodily functions.*

Further to this, one justification for adopting IPA as the main analytical technique is that this study is heavily reliant on the utilisation of a double and sometimes triple hermeneutic: “the participant is trying to make sense of their personal and social world; the researcher is trying to make sense of the participant trying to make sense of their personal and social world” (Smith, 2004: p.40). The hermeneutic turn in IPA complements the multiperspectival approach that this thesis adopted in developing a narrative about the relational experiences of living with NPC, whether from the person diagnosed or their family carer. The utilisation of IPA means differing personal perspectives on the world can be understood by a third party by focusing on patterns of meaning-making, illuminating the shared experience of living with NPC and the interaction between lifeworlds. This was particularly important in this study as many of the people diagnosed with NPC were unable to take part in an interview by themselves, if at all. Via the process of parents and carers being asked to ‘put themselves in the shoes’ of the person they were caring for, I was able to glimpse the meaning of the illness experience for both people living with NPC and their carers. Larkin et al. (2018) suggested that in multiperspectival IPA, the researcher is a “sort of ‘meta-phenomenologist’”, re-interpreting all of the participants’ individual interpretations (p.14). With this said, the difficulty of using first and second-hand accounts attempt to engage with the participant’s personal and social world is recognised whilst maintaining the belief that the objective of IPA is to acquire descriptions which get as ‘close’ to the view of the participant as is possible (Larkin, Watts, & Clifton, 2006).

The concept of the hermeneutic circle within IPA represents an iterative process as opposed to the following of linear steps. The back and forth motions within the analytical process are performed in order to grasp the parts, viewing the text from a variety of vantage points to provide a considered and in-depth representation of the phenomenon being explored (Smith et al., 2009). Larkin et al. (2018) argued that this need not involve the loss of idiography by looking at multiple perspectives, as long as the analyst is sensitive to these risks. Smith et al. (2009) argued that “when people are engaged with ‘an experience’ of something major in their lives, they begin to reflect on the significance of what is happening and IPA research aims to engage with these reflections” (3). This complemented the main aim of the illness narratives that were being engaged with.

It was for these reasons that IPA was chosen over other qualitative analyses. IPA worked in harmony with the research paradigm of this thesis and was therefore better-suited than thematic analysis, which does not stem from a particular epistemological or theoretical position (Braun & Clarke, 2006). The epistemological emphasis on the interpretative stance of this thesis meant that the lived experience was of central focus as opposed to the more general goal of thematic analysis in identifying themes. IPA honoured the ontological stance required for gaining understanding of the experience of illness, and the existential nature of life and death that was being explored.

IPA was deemed more suitable than grounded theory due to the central focus of grounded theory being to produce a theory or a number of theories from its utilisation (Willig, 2008). This did not fit the aims of this thesis, as I wanted to explore experiences as opposed to generate theory. In addition to this, grounded theory suggests that the researcher acts as an observer who is able to portray a representation in the analysis without this being based on subjective assumptions or expectations (Willig, 2008). As spoken about before, my epistemological position is that data collection and analysis is influenced by the researcher's experiences and presumptions and thus cannot be separated in this manner. According to Smith et al. (2009), IPA was developed as a method that was focused on the meaning-making of people's experiences as this was something that other qualitative methods had not prioritised. In order to elucidate the experiences of living with and dying from NPC, IPA was therefore felt most appropriate for the current endeavour.

The adoption of IPA in this study complements the biographical data collection method, as IPA is well-matched to an approach where participants are invited to give a detailed account of their experiences (Smith et al., 2009: p.56). In addition to this, this study did not seek to recruit large numbers of people with NPC to interview. Firstly, this would not have been possible due to NPC being a rare disease. Secondly, when using IPA with a small sample, the idiographic commitment IPA endorses is not lost (Noon, 2018). Smaller samples for each group, i.e. children with NPC, adults with NPC and bereaved parents allowed me to commit to a thorough and in-depth analysis of each case, which in turn enabled me to concentrate on the idiosyncrasies of particular experiences (Noon, 2018). IPA enabled a biographic account of the meaning-making of experiences and acted as a springboard for understanding the phenomenon of living with a rare disease more widely.

Finally, another reason for adopting IPA as the main analytical technique was its commitment to reflexivity. Goldspink and Engward (2018) argued that when reflexivity is used in IPA it not only strengthens the rigour of the research process, but enables the researcher to achieve an interpretation that has greater depth when analysing the data. In this thesis, combining IPA and reflexivity in dialogue with the lifeworld theory provided a multidimensional approach to interpretative work in understanding living with incurable illness.

3.7.2 Embodied Interpretation

Embodied interpretation as an analytical technique is a “body-based hermeneutics in which qualitative meanings are pursued by a back-and-forth movement between words and their felt complexity in the lived body” (Todres & Galvin, 2008: p.575). This analytical method is situated within an interpretive epistemology, which is motivated by an aesthetic portrayal of phenomenology. Its focus is situated on the evocative use of language in meanings, sharing participant’s spoken words in a more ‘felt’ meaningful way (Todres & Galvin, 2008). Todres and Galvin (2008) argued that an interview is more than just the verbal expressions participants speak and ‘the language of the body’ should be acknowledged within the interview and beyond this, in the analysis. The researcher should recognise or be mindful of both spoken and unspoken language as they both express meaning of the participant’s world. Finlay (2006) spoke about this as the body disclosing the world to the researcher, for example through gesture and demeanour, as opposed to phenomenology purely relying on words spoken and transcripts. As Kvigne et al. (2002) articulate:

> *“The language of the body may both reinforce and weaken what is said, or may even communicate a message at odds with what is expressed verbally. Noting for example the body’s self-representation with regard to appearance and the care taken with one’s appearance, movement and function can form a basis for questions and thus for greater depth in the informant’s own descriptions” (p.65).*

Embodied interpretation was utilised in the third empirical study of this thesis, which looked at end-of-life care and the dying experience of a child to NPC from the bereaved parent’s perspective. Exploring the end-of-life and the dying experience called for an analytical approach that was more than just interpretation, but one that tapped into the ‘felt sense’ to help steer the understanding of these experiences. This was due to wanting to gain a more fulsome and holistic appreciation of one of the most difficult experiences one can face in life. Due to the sensitive and raw experiences of losing a child, both words and the bodily expressions needed to be paid attention to as the ‘whole’ as participants walked me through the last few moments of their child’s life. During and mostly after conducting the interviews, I would write notes formed on my intuition and felt-sense of being with the participant, being a part of their world as they shared their most vulnerable times. The notion of felt-sense refers to going beyond language alone, communicating the experience and interpretation of the individual beyond the words spoken. Todres and Galvin (2008) argued that these notes taken and used in analysis are meaningful and provide an interpretation of the situation as the researcher interacts and interconnects with the intersubjective world; understanding the body’s language of expression. Using my felt-sense to develop depth and meaning in both the interview process and the data analysis process necessitated a practice of ‘indwelling’, or turning inward to understand the nature of human experience when approaching the end of life (Moustakas, 1990; Pascal, 2010). Thus, the application of embodied interpretation seeks to understand more than just abstract cognition (Amos, 2016). As the bodily conduit for the experience imparted by the participant, the researcher has a role to play in shaping the narrative and its meaning (Finlay, 2006).

The analytical approach used to give an embodied interpretation, follows Finlay's (2006) framework, where she suggests three stages that should be attended to when applying embodied interpretation in phenomenological analysis. These will be discussed in close detail in the methods section in chapter 6. These stages: Bodily empathy; Embodied self-awareness; Embodied intersubjectivity are wrapped up within the importance of the continuation of reflexivity. This strategy was particularly emphasised in how the results of this chapter are written, with the embodied interpretation being used to add a further layer of continued reflection to the meanings that have been drawn out. The process of immersion in the data of these experiences was at times upsetting for me. But, I believe that channelling this felt bodily empathy enhanced my reasoning and interpretation of the analysis.

This practice of using my intuition together with the data and the dwelling-mobility lattices provided an in-depth and empathic understanding of the last moments of life and the dying experience. This creative and iterative approach offered a more humanised perspective to see and to feel the evolving story. Following and reinforcing Merleau-Ponty's (1960/1964) belief that "It is through his body that the other person's soul is in my eyes" (p.172), this analysis was a union between words and my observations with the undercurrent of the existential theories of suffering and well-being carrying me, portraying an aesthetic phenomenology of death and dying.

3.8 Ethical considerations

The sensitive nature of this study called for a rigorous approach to ethical considerations that needed to be put in place to protect the participants and myself. Concerns relating to ensuring the understanding of informed consent, confidentiality, and privacy are of high importance and were adhered to in this study. These will now be discussed.

The study sought and was granted NHS ethical approval (ethics number: 15/WM/0093) (see Appendix 1) to enable people to be recruited from NPC clinics and from the NPUK charity. Due to the rarity of the condition and the links the NPUK charity have with the NPC community, all participants were recruited via the charity. Mid-way through my PhD, I submitted an ethics application to the Aston University Ethics Committee for the exploration of end-of-life and the death of a family member from NPC study, which was given a favourable opinion (project ♯1259) (see Appendix 2).

Potential participants were given information sheets (see Appendix 3 and 4), which outlined the purposes of the studies and what would be involved in taking part. These described the studies in sufficient detail and highlighted any potential risks of taking part, which were mainly surrounding the idea that both people with NPC and their families may find talking about their experiences of living with and dying from NPC distressing. I was concerned about participants experiencing distress due to the personal and sensitive nature of talking about an incurable illness and in some cases, the death of a loved one to NPC. Participants were reminded several times throughout the entirety of their participation (on the information sheet, consent form and during the interview) that they could terminate the interview at any given point, without having to give a reason for doing so. They could skip any interview questions or could take a break from the interview at any point. I went through the consent forms (see Appendix 5 and 6) prior to starting the interview. This was to ensure participants were happy with what was involved before they gave their consent. Participants with NPC who were under the age of 16 years who took part in the study needed parental ascent to be given before they could take part themselves in an interview.

Participants were told that they could withdraw from the study at any time during the interview without giving reason for doing so. They were also made aware that they could withdraw from the study at any point during the 2 weeks after being interviewed and that following this, the data from the interview would be included in the analysis. Participants were given a code after their interview and were told to quote this when contacting the researcher for any questions following the interview or if they wanted to withdraw from the study. Participants were made aware that this study was separate from the medical care they were receiving and that this would not be affected if they chose to withdraw.

Throughout the entirety of the study, I was in close communication with the NPUK charity, informing them of the interviews I had conducted. A couple of days after each interview, g, NPUK would make contact with the participant to enquire as to their welfare. Participants were aware that NPUK would be informed of their participation and would be in touch following the interview. Participants were given contact details of the research team and encouraged to contact the team to discuss questions or to withdraw participation if they so desired. I was sometimes joined by the NPUK nurse at interviews, especially if the NPUK charity thought that the participant would benefit from having a familiar face present during the interview. All interviews were recorded and transcribed anonymously and saved to a password-protected computer. All demographic information that was needed was taken during the interview, such as gender and age of person with NPC and how long they had been diagnosed for.

One important ethical consideration was the potential of participants being identified and I was particularly concerned about confidentiality. Due to the rarity of NPC, there was a possibility that participants may have been identifiable to people in the NPC community within the dissemination of the studies' findings. To protect participants, names of people and places were altered to pseudonyms during transcription, and all data were de-identified: no participant details were stored within the transcripts. Data cleaning occurred for each interview, where the researcher removed any identifiers both on a personal and contextual level for each participant. For the two empirical studies looking at the experiences of children and the experiences of adults living with NPC, data from the participants were merged together to create new characters. This will be spoken about more in each empirical study, but on the whole was done by merging together characters who displayed similar progression of symptoms. For example merging participants who had similar deterioration in terms of mobility or merging participants who were of a similar age. This was carried out in order to protect the identities of those involved whilst ensuring that the individual experiences could still be conveyed. Although there was a slightly increased risk of individuals being identifiable within the NPC community, the need to hear the distinct 'voices' within differing accounts was judged to be significant enough to justify this action. Very little in respondents' quotations was altered, ensuring the authenticity of the data collected. Arguably the idiographic commitment of IPA is challenged by the data handling decisions of this study but great care was taken to protect the subjective experiences of the participants.

On the contrary, the study looking at the dying experience, spoken about in chapter 6, section 6.1.4, the method of data cleaning was not as prescriptive for the end-of-life and dying experience study. Pseudonyms were given to protect the participants' identities but I felt that altering gender or ages for this study would take away the rawness of the narratives that the families were conveying. The risk of identification within the NPC community was explained to the participants due to the closeness of the community and the sharing of experiences. All participants understood and agreed to this risk. One motivation for taking part in this study was the hope that their openness would benefit others who had lost a child to NPC, thus they wanted the narratives portrayed to be as authentic as possible.

Acting with ethical integrity goes further than those which committees and national guidelines set out (Ezzy, 2002; Kellehear, 1993). As Pascal (2010) wrote: "There is a personal and moral obligation on the researcher to treat participants with respect for their knowledge, experience and human rights.” (p.11). This formed and guided the ethical considerations overall, seeking to respect and honour the participants’ stories.

3.9 Summary

The methodological approaches used in this thesis are deemed the most suitable to explore in depth the lived experiences of NPC within the interpretivist paradigm which directed these decisions. Engaging in an explanation-driven logic of inquiry enabled a way of understanding the experiences of living with and dying from NPC enhanced by the use of lifeworld theory. This strong reflexive focus enabled the series of studies to uncover new meanings of living with NPC from people who have been impacted the most.

The series of studies are as follows. Chapter 4, constitutes an understanding of the experiences of children living with NPC through their parents’ sense-making of their children’s experiences; and secondly to this, to make sense of the parents’ own experiences of caring for their child with NPC. Chapter 5 will explore adults’ experiences of living with NPC. In this empirical study, we hear more from the person diagnosed with NPC compared to the previous chapter. Family carers’ voices are still present and the ways in which their identity has changed from ‘parent’ or ‘spouse’ to ‘carer’ is explored. Chapter 6 explores the final days of living with NPC from the perspective of two families whose child had died. End-of-life care is discussed and how this impacted upon the dying experiences is conveyed. An exploration of how life is conceptualised after death occurred is also included.

In order to tie in with the original aims of the study, which were to develop and validate QoL scales for people living with NPC, chapter 7 presents the development of two QoL scales for NPC and includes a reflexive account, which uses the richness of data acquired from the previous studies to suggest a novel approach to scale development grounded in phenomenology. Quite often in phenomenological research, powerful insights are gained through the in-depth experiential perspectives, which have been illuminated, yet the question of how these important findings can be used to inform clinical practice or policy is left unattended (Galvin et al., 2018). This chapter will demonstrate a methodological process of developing items for a QoL tool that could be used in a clinical setting formed on the existential meanings of living with NPC. Attention will be given to the use of a tool such as this within a clinical setting and its potential for humanising care within a rare disease community. The findings from this thesis will be synthesised and discussed in chapter 8, which is the final discussion chapter.

Chapter 4 - "I do sometimes just think she looks so fed up." A multiperspectival approach to understanding the accounts of children living with NPC.

4.0 Introduction

Illness has been framed as an experience of "uncanniness", an obtrusive invasion on life as we know it (Carel, 2013: p.249; Svenaeus, 2000c). Svenaeus (2011) likened Heidegger's explorations of suffering, the events that "break in on us and destroy us," to being diagnosed with severe illness (p.335). For Heidegger, who was not precisely concerned with illness, these events were referred to as objective experiences, such as natural catastrophes including earthquakes or tsunamis (Svenaeus, 2011). These natural disasters are unmeaningful (*unsinnig*) and absurd (*widersinnig*) (Heidegger, 1962: p.151-152) in that they "resist meaning" in their completely alien nature which serves to overwhelm our previously held sense of what it means to *be* (Svenaeus, 2011: p.335). Svenaeus (2011) suggests that this forced unfamiliarity as to how we exist in the world could be the same when one is diagnosed and lives with severe disease; it "breaks in on us and destroys us" (p.335) in a similar way. The relative inexperience of this way of *being* means that an individual can struggle to generate a meaningful sense of life's narrative and self-understanding. When forced beyond a frame of reference for meaningful existence, the individual can be left cut-adrift and overwhelmed. This framing of the concept of illness is given further clarity by the suggestion that a Heideggerian sense of at-homeness can be revoked by the illness experience. The individual can be described as not feeling at home in the world, or in their body (Moran, 2000; Toombs, 1993). When we are well, we feel at-home in our bodies, but when illness enters our world, we begin to feel "disharmony, dis-equilibrium, dis-ability and dis-ease which incorporates a loss of the familiar world" (Toombs, 1993: p.96). Svenaeus (2011) articulates it well: "Illness is an unhomelike being-in-the-world in which the embodied ways of being-in of the self (person) have been thwarted." (p.337).

Carel (2013) suggested that it was possible for one to feel at-home within illness and subsequently experience well-being. Thus, living with illness does not necessarily mean a permanent negative disruption and instead could be seen as something that could bring positive consequences to the lives of those living with it. Living with illness can lead to self-discovery and self-development (Charmaz, 1983; Moch, 1989). Carel (2007) argued that although illness can generate a division between the objective and biological on the one hand, and the subjective lived body on the other, reconciliation is achievable by giving precedence to adaptability and “creative responses to illness and disability” (p.108). Galvin and Todres (2011) suggested that we can feel a sense of at-homeness in a given situation through our sense of rootedness and *dwelling* and that this ‘letting be’ can be congruent to a peaceful sense of being. With this said, whilst it should be recognised that illness is a universal and a significant element of being, the experiences of illness are as varied as the people who encounter it (Carel, 2013). It is therefore important to recognise the co-existence of being at-home and homelessness when living with a LLC in order to understand in more detail the lived experiences of this.

Within the context of children being diagnosed with and living with incurable illness, research has focused on the experiences of caring from the perspectives of parents and siblings. As seen in chapter 2, there appears to be a paucity of research that has sought to understand from the child’s perspective how they make sense of living with a LLC. When looking at this in relation to this study, the manifestations of NPC means it is sometimes difficult to fully understand how children living with the disease experience their illness as they may find it difficult to communicate their thoughts with clarity and accuracy. This gap in the wider research of LLCs is perhaps exacerbated by the ethical dilemmas of accessing such a perspective, along with the nature of the illness creating barriers to such an understanding. With this said, Merleau-Ponty (1962) sought to show that perception should not be reduced to a neurological process, but should be understood as the body as a whole and its relationship to its environment. It is reductive to ignore that our sensory and physical relationship to the world precedes our perception of it in any meaningful, individual sense. Taking this bodily, and indeed *embodied*, understanding of everyday experience and perception, allows for an important perspective on our encounter of being-in-the-world (van Manen, 2012). In his research exploring the experiences of parents in the neonatal hospital setting, van Manen (2012) argued that: “We need to attend to the question of how the infant’s preconscious perceptual systems (hearing, sensing, smelling, and feeling) are subtly but complexly integrated into the physical environment in which the child is placed. Yet, at the same time, we need to gain a more differentiated understanding of how, at this prereflective level, the parents perceive their infant’s situated-ness and subjectiveness to the care.” (p.201). There is, within this argument, a recognition that even when the perception of a cared-for individual is limited, there is still value in the interpretation of an individual’s experience of being, especially by those closest to them. Thus calling on multiple experiences to gain understanding of the phenomena in question is both possible and useful, providing an important precedent for the aims of this present study.

This leads to the claim that it is important for research to seek understanding in how children diagnosed with a LLC make sense of their experience and the impact it has on their daily life. Crucially, this would seek to transcend the objective limitations of barriers to communication, which traditional methods would discount as inaccessible. In order to address the emerging research needs in this area, it is necessary to construct a framework of inquiry that adequately illuminates the issue whilst mitigating for the potential ethical and practical challenges. One way of achieving such a goal is to build on an understanding how people living with illness make sense of their experiences from a multiperspectival approach. This can be pursued via the phenomenological hermeneutic approach outlined in chapter 3.

When a child is diagnosed with a LLC, it can be characterised as a disturbance (Bury, 1982) to the narrative framework of the whole family, a significant threat to the narrative constructed by the family members and the goals for family life they had imagined (Reeve, Lloyd-Williams, Payne, & Dowrick, 2010). The lifeworlds, the context in which we experience and make sense of the world, within a family are in close connection and the family unit share the experiences of being-together-in-the-world in the lived experiences of daily life. Through this relationality, the inherent, interpersonal connectedness that characterises family life is impacted by the illness experience. Beyond this, the interconnected lifeworlds of the family can also provide insight into the experiences and meanings of individuals whose perspective it would ordinarily be difficult to access; where the lifeworld of one family member can be used as conduit for the viewpoint of another. The lifeworld is therefore a meaningful theoretical position from which to explore these specific experiences of life with incurable illness. Drawing on the interrelated elements of the lifeworld - temporality, spatiality, intersubjectivity, mood, identity and embodiment (Todres & Galvin, 2006) - we can further explore how this can be changed or disrupted, by living with illness. Therefore the aims of this study are to understand the experiences of children living with NPC through their parents' sense-making of their children's experiences. Secondly to this, it aims to make sense of parents' own experiences of caring for their child living with NPC in order to understand more about the disruptions to the lifeworld for close family carers.

4.1 Method

4.1.2 Study Design

An exploratory study was chosen which was rooted in phenomenology. The phenomenon in this study is living with NPC as experienced by parents and children. The inductive method, which is driven by data descriptions derived from individuals led the focus of the analysis. In addition to adopting this inductive approach, the existential theories of well-being and suffering were drawn upon, thus evoking an abductive logic of inquiry (Hiles, 2012). This study received a favourable opinion from the Research Ethics Committee (ethics number 15/WM/093).

4.1.3 Participants

Seven parents of children with NPC participated in the study, all of whom were recruited from the NPUK charity. These were five mothers and two fathers. All participants had a child aged 0-12 years who had been diagnosed with NPC (see Table 4.1). Participants were recruited purposively and all were UK residents. Following discussions with parents, all decided they did not wish for their child to be directly involved in the interview due to either the cognitive manifestations of NPC in the child diagnosed, the child's age or the general concern that the interview would not be beneficial for the child. Parents of children with NPC were therefore asked to be part of the interview on behalf of their child. These were conducted face-to-face in the home of the participant (n= 3) or over the telephone (n = 4) according to each participant's preference.

4.1.4 Data collection

An advertisement for the study was published on the NPUK Charity's social media pages and interested participants contacted the researcher. Written consent was taken from parents prior to interview. The interview schedule comprised of open-ended questions, which focused on the child's NPC story (see Appendix 7). Firstly, parents were asked to talk about their child's experiences. The researcher asked parents to put themselves 'in their child's shoes'. Questions then turned to their own experiences of caring for a child with NPC. Interviews were recorded using a dictaphone.

4.1.5 Analysis

The interview data were analysed using IPA (Smith et al., 2009). As outlined in chapter 3.7.1, IPA enabled a biographic account of living with NPC. For this study, this involved a triple hermeneutic, in that I was understanding the parents' meaning-making of their child's experiences. Guidelines provided by Smith et al. (2009) were followed, however this was not purely a linear process but was iterative and flexible in its nature (Pietkiewicz & Smith, 2012). The analysis process is fluid and multi-directional but I have outlined the general steps taken in table 4.2.

Following each interview but prior to transcribing, I spent time writing notes about how the interview had gone, what I could do better next time, what additional questions I could have asked and the participants' story. This would be used reflectively in thinking about my analysis, especially with regards to considering non-verbal communication and the mood of the interview i.e. whether the interview had felt rushed or if the participant's body language had changed throughout the interview. This follows Shaw's (2010) suggestion of writing a summary of the person's account to better understand the nuances of the interview.

4.1.5.1 Transcription

All interviews were transcribed verbatim and this process was a key part of becoming familiar with the data. Table 4.3 denotes the transcription notations, which were used throughout transcribing. Audio recordings were listened to a second time to become more acquainted with the data and to check the transcripts were appropriate representations of participants'accounts. This helped in immersing myself into the data. All transcripts were printed to be annotated by hand.

4.1.5.2 Reading and note making

This stage involved reading each transcript through several times to provide familiarisation with the data and to get as 'close' to the phenomenon as possible. During the first read through, free coding was utilised, maintaining an open mind and focusing on the initial ideas that emerged. This included descriptive comments and interesting linguistic contents used by the participant such as: key relationships, places, significant appointments emotional responses, pauses, the degree of fluency and repetition (see Appendix 8 for an example of my annotated transcripts). Detailed notes and comments on the data to differentiate between descriptive, semantic and conceptual comments was conducted using different coloured pens.

As Smith et al. (2009) suggest: "it is important to engage in analytic dialogue with each line of transcript, asking questions of what the word, phrase, sentence means to you, and attempting to check what it means for the participant." (p.84). This was a time-consuming process but also a very detailed way of immersing myself into the participant's lived world. It was also emotionally difficult at times, as I straddled my own experiences with those of the participants and considered the magnitude of that which the participants were sharing, opening up a range of provisional meanings.

Table 3. General stages in the IPA analysis - adapted from Smith et al. (2009).

	Brief description	**Purpose**
Step 1	Transcribing	To gain familiarity with the data by listening to the participant's stories and create for analysis.
Step 2	Reading through transcripts	To immerse oneself into the participant's world.
Step 3	Free coding and exploratory discussion	To focus on interesting language both descriptive and linguistic and produce detailed notes on what appears meaningful to the participant's world.
Step 4	Developing emergent themes	Mapping patterns and connections between exploratory notes.
Step 5	Searching for connections	Developing super-ordinate themes that appear to categorise the most meaningful understandings of the phenomenon in question. Steps 1-5 to be repeated again for each case.
Step 6	Searching for patterns across cases	Explore super-ordinate themes for all participants and illuminate the similarities and differences between the meanings attached by the participants.

4.1.5.3 Generating themes

From the comprehensively annotated notes, I began to develop emergent themes. This comprised of mapping connections and interrelations between the exploratory notes produced. This involved breaking up the narrative flow in order to generate initial themes. These fragmented parts started to reflect the essence of what was unearthed from the transcript, as I began to produce short and meaningful descriptions of what was important in the participant's story. A list was produced of all emerging themes, including those that were repeated, as well as themes that were worded differently but had similar meanings.

4.1.5.4 Connecting themes generated

Themes were grouped together, some under existing theme names, others under new names. This process involved the clustering of initial themes forming sub-themes. Some themes were discarded due to there not being enough evidence to retain them. This process of refining the information available drew together the ideas that revealed the most meaningful elements of life with NPC for the participants and their families. Practically, this was achieved by colour coding the emerging themes throughout a Word document to allow for connections to be made across the data. This is where abstraction took place and the cluster of emerging themes could be grouped together to form a super-ordinate theme, those being concepts, holding hierarchical priority over other ones. Subsumption also took place for one emerging theme, which acquired a "super-ordinate status" (Smith et al., 2009: p.97).

4.1.5.5. Continuing to the next case

This process was repeated for each participant. Smith et al. (2009) highlight the importance of treating each participant's story as a case in its own right, thus I needed to bracket as much as possible, the emerging ideas from the previous participant and the stage of analysis that I had got to, in making sure new ideas and new key themes could emerge.

4.1.5.6 Looking for patterns

To discover the overall master themes I explored the connections between individual participant's themes by writing down each super-ordinate theme on a large piece of paper and mapping connections and making notes of what each theme represented. Smith and Osborn (2003) make the important point that one of the ways in which the selection of themes should be determined is by the richness of the idea and its illumination of the account as a whole, rather than merely how often it occurs in the data. As such, theme selection was based on the pertinence of the phenomenon being discussed, but also its significance to one or two participants in order to preserve the idiographic nature of IPA analysis. This stage also sought to incorporate the elements in the lifeworld, using this theoretical angle to draw connections between themes.

Planning the structure of the written analysis was key before beginning the writing process. I sought to structure my analysis with themes and experiential accounts, in such a way as to illustrate the nuances of the narrative as a whole. Galvin and Todres' (2013) existential theories of well-being and suffering were used to help make sense of the inductive themes arising from the IPA analysis. These existential theories were adopted because of their commitment to describing and understanding the lifeworld in its wholeness, as a set of interrelated dimensions (Todres et al., 2006) - see Tables: 3.1 & 3.2 for the dwelling-mobility lattices. This process involved a cyclical dialogue between the themes generated alongside the existing theoretical concepts, thus grounding the inductive analysis. This open dialogue between each theme and each element of the lifeworld led to exploratory notes being taken of how the themes and elements interrelated. This enabled a more fulsome exploration of the themes against the constituent parts of the lifeworld. This process helped in structuring my analysis, particularly when a certain element of the lifeworld was found to be relevant and/or illuminating of other concepts within the analysis. This remained an iterative process, moving between the transcripts and listening back to the interview recordings to immerse myself again in the account rendered. In addition to these broad methodological steps, the analytic process was discussed with my supervisors.

4.1.6 Data handling

Due to the rarity of NPC, there was a possibility that participants may have been identifiable to people in the NPC community through the dissemination of the study's findings. Data cleaning was carried out for each interview, where any identifiers both on a personal and contextual level were removed. A further measure to protect the identities of the participants was pursued through the merging of participant data to create three new characters (Hopkins, 1993; Saunders, Kitzinger, & Kitzinger, 2015). This was achieved by combining the narratives of participants who displayed similar progression of symptoms. For example two participants with similar deterioration in terms of mobility were used in one composite, and others were grouped due to very little physical deterioration. In order to avoid losing the richness of personal experiences, amendments were made to information deemed non-essential for this study such as gender, ages and family details. Very few respondents' quotations were altered, ensuring the authenticity of accounts presented.

Table 4. Transcription notation

I:	Interviewer
P:	Participant
M:	Mother of someone with NPC
F:	Father of someone with NPC
…	Indicates a short pause
[pause]	Indicates a longer pause
***	Represents an inaudible comment
Capital letters	Used to denote emphasis
[ ]	Short interjections made whilst someone was talking e.g. [I: okay]. Used to add clarity to certain quotes, such as who the participant was referring to, or the name of a place a participant was talking about. It was also used to indicate non-verbal or emotion descriptions such as, [starts to cry]

4.2 Results

4.2.1 Pen portraits

Jane: Glen's daughter, Jane had been diagnosed as a baby with NPC. The progression of NPC in Jane happened quickly in the last 18 months.

Felicity: Diana's daughter, Felicity, had been diagnosed when she was three years old. Felicity was well physically and lived an active life.

Elliott: Stephanie's son, Elliott, was diagnosed when he was five years old. He had declined rapidly over recent years. See table 4.1 for participant information.

Table 5. Participants

Name of child	Name of parent	Age of child at diagnosis	Age of child now
Jane	Glen	4 weeks	8 years
Elliott	Stephanie	5 years	10 years
Felicity	Diana	3 years	7 years

The themes identified were: *Spatial and temporal loss, Relationships as security and enablement* and *Creating opportunity in the face of adversity.*

4.2.2 Spatial and temporal loss

The manifestations NPC was having on the objective body was in most cases the starting point for parents' perceiving how their child made sense of living with illness. Attempts to convey meaning began with what was observable and noticeable; what the observed body can no longer do compared to what it once could. Although there were similarities described by all parents about the manifestations on the objective body, the differing level of physical deterioration between the children was noticed when analysing the data, and this was co-existent with the extent to which issues were discussed. Glen spoke extensively about the ways in which his daughter had deteriorated.

> Glen*: "…she can't sit up anymore by herself you know… she's in danger of kind of toppling over and hitting her head on something… even in her bed… you know she used to get up and crawl around her bed… but… yeah she can't … she can't move now she can't even turn over herself…"*

Glen's comments were indicative of the constant care that Jane required, reminiscent of the care a baby needs. This represents a revealing example of the embodied connection between carer and cared-for, the inextricable connection of the lifeworlds. The particular type of care a baby needs is dependent on the attention and attentiveness of others to facilitate a safe and content relationship with the world. Emphasis was given to how Jane would need such care even in the relative safe environment of her bed. This suggested that Jane experienced a lack of positive freedom in the traditional sense of freedom 'to do', a concept exacerbated by a temporal and spatial understanding that is characterised by dependence and limitation. Time cannot be spent alone, restful spaces are restrictive and any comfort is dependent on others.

This evoked feelings of frustration in Jane, shown mainly through facial reactions due to her inability to communicate.

> Glen: *"but yeah I do sometimes just think she looks so fed up [yeah]."*

The element of mood here is key in transcending the objective body into the lived body, and indeed the hermeneutical connection between the child, the carer and the researcher. The language of Jane's mood spoke straight to Glen in her weariness of the illness experience, which he was then able to interpret and convey. This mood did not manifest in an agitated appearance but in a more closed-in attitude, restricted to her wheelchair with limited horizons. There are times when a lack of energy and motivation were evident in Jane. As Glen's story progressed he spoke at length about how he understood Jane to experience life with NPC with a degree of contentment and was able to generally express herself as a *"happy girl".* As a caveat to this, Glen needed to further explain that despite his daughter's cognitive decline, he felt that Jane understood and recognised that she was living with illness and he suggested that Jane experienced this by eliciting feelings of depression.

> Glen: *"... I I think most of the time she's quite happy... but I think she understands probably… that she can't do things anymore... you know it's it's almost as if she's got to a point where she's stopped developing and life is working its way back down to baby stages."*

A sense of entrapment relating to Jane's embodiment is expressed here. The objective body will not allow her to do what her selfhood desires, that which has been the basis of her identity in the past, one in which she was physically and communicatively able. This sense of a lost future, a loss of potential, was also reflected in her mood. There is a sense of resignation to the deterioration being experienced, a sense of being increasingly alienated from the body she inhabited. Glen can sense this frustration in Jane within her interactions; even though words are decreasing, there is still meaning being conveyed between them.

> Glen: *"I remember crying… you know… at her frustration and I felt so helpless… you know I couldn't do anything [yeah yeah] just take care of her but she does get very frustrated…"*
>
> I: *"Why do you think she felt frustrated?"*
>
> Glen: *"yeah ... sorry ... I mean her decline has been so much so quick she can't even lift her arms, [mmmm] you know... she just needs somebody to do everything for her"*

Here we see the expression of a different aspect of Jane's experience than had previously been intimated, one characterised by frustration and agitation instead of resignation or by being subdued. The rapid decline is cited as the catalyst for a sense of entrapment following the loss of physical capability. This loss can be thought of as the objective body becoming alien to Jane's lived experience, where agitation and restlessness have come to dominate. It was also telling to hear Glen speak of helplessness, a kind of admission of being pushed beyond the limits of parental care. This helplessness met Jane's frustration in a merging of the lived experience where the lifeworlds of both were shaped and moulded by the other. The security of familial relationships was challenged by the deterioration brought on by illness.

Stephanie spoke specifically about Elliott's lack of awareness of his deterioration, which was described as being positive for him. Stephanie's reaction could be understood to be typical of a parent in a number of ways. Firstly, she wanted to believe that this lack of awareness was the best outcome for her son. Secondly, Stephanie wanted to protect Elliott from harm in a maternal sense. These reactions reveal little of her son's understanding of illness.

> Stephanie: *"I mean you know it fortunately… Elliott's affected more more neurologically than anything else [yeah] you know which sounds horrible but … He doesn't realise what… what's goings on these days…"*

This use of the word "fortunately" here denotes that this cognitive impairment is considered a good thing for Elliott; an outcome that allowed him to better cope with the realities of his illness. Stephanie interpreted Elliott's experiences as being characterised by being in his own world, a world that is free from frustration and agitation. Stephanie's emphasis on the impact and peculiar mercy of Elliott's neurological limitations does not stop her from recognising the decline in physical mobility and the social consequences of this.

> Stephanie: *"…at parties he would normally get up and have a dance whereas he's not going to be able to… [starts to cry] sorry it's then you realise… When you're on a day to day basis... you kind a don't realise … how things have changed."*

The concepts of temporality and spatiality seemed to be lenses of experience for both Stephanie and Elliott. Comparing her son's physical decline to how he was a year ago emphasised the prominence of deterioration. From Stephanie's perspective, decline was obvious, especially when comparing the changes over a longer time period. This temporal element of the lifeworld is a significant way of understanding how Stephanie makes sense of his illness in that she compared what he *was* like then to what he *is* like now. The alienation of Elliot from his peer group's experiences, and from his mother's prior reflections on what he might have been, creates a sense of dissonance between expectation and reality that speaks to a deeper need for communal experience and future orientation. This had emotional ramifications for Stephanie, and a sense of permanent loss was expressed. Yet, this is not communicated in how she felt her son made sense of it.

> Stephanie: *"…it's pretty crap it's almost as if he's got to a point where he's stopped developing and life is working its way back down to baby stages [yeah]…but again nothing bothers him really"*

From Stephanie's temporal view, there had been a halt in the progression of life, it was no longer developing in a conventional chronology, instead moving backwards towards a son that was once again helpless. Yet, Stephanie commented that her son had taken this in his stride and that there was a hint of him moving towards his own sense of space. Stephanie questioned whether Elliott now showed less frustration due to the cognitive manifestations of NPC or whether there had now been a level of acceptance, with the indicator of the potential acceptance being in the way Elliott now showed less frustration.

> Stephanie: *"he gets frustrated and agitated but again I think that's more … … maybe not so much but when he first started getting agitated when he was losing mobility I think that was more down to knowing that he could do it before but now he can't… again you know whether it's the disease or acceptance but he's kind of fine with it now"*

Although moments of frustration were suggested, there still seemed to exist a reconciliation of his body to its limitations, a spatial understanding uncoupled from traditional measures of contentment. There is a sense in which a self-actualising understanding of one's own temporal narrative is dependent on a certain level of cognition. Without this, the pressures of comparison and achievement do not exist; one is freed from common understandings of happiness or success. Comparatively, Jane appears to express this more frequently than Elliott. It must be noted, however, that both these examples are dependent on the interpretation of the caregiver. Whilst, as discussed above, this can be an important contributor to our understanding of the child's being-in-the-world, one must not fail to recognise the influence of the parent's lifeworld and self-narrative on the perspective offered.

Diana's daughter Felicity was more physically able than both Elliott and Jane. Diana commented on how her daughter had been experiencing mood swings and commonly presented frustration.

> Diana: *"…because she has a problem with understanding what goes on around [her] sometimes… Mood swings are very much up and down so that you can definitely tell that something is wrong"*

Diana explained how her daughter would get frustrated when she could not do the same things as her friends, emphasising the meanings her daughter placed on friendships.

> Diana: *"she's aware that she's different in she needs help with things but the others in her class they can do it by themselves and that frustrates her because she wants to do it by herself as well… she gets Very frustrated when she can't do things... when she's not able to say things."*

A lack of freedom is illustrated here; there is an emotional reaction noted to a felt sense of entrapment, in her identity as one limited by illness. Diana's daughter displayed many frustrations: firstly that of being different to others and; secondly that of being dependent upon others with the latter seeming to cause the most disruption to Felicity's sense of self. This comparison that Diana suggested between Felicity and her peers is echoed in the framework of symbolic interactionism, the relationship between self and society. The subjective meanings that Felicity placed on her interactions and the comparisons that she made between her 'self' and her peers, created the meaning she gives herself. The interconnectedness between identity, intersubjectivity and embodiment is demonstrated through feelings of frustration with what her body could not do compared to what others could. The sense of being alienated from her hoped-for self and from those around her therefore became apparent in this intersubjective space.

The bodily changes and diassociation this had on body-self signaled that the body can no longer be taken-for-granted. As Carel (2013) articulated: "the body thwarts plans, impedes choices, renders actions impossible" (p.247). The body as other was perceived by all children.

4.2.3 Relationships as security and enablement

Parents spoke about the importance of family, especially siblings, to their children living with NPC. Siblings were described as bringing a sense of enjoyment to their child's life, and in some regard, acting as a distraction.

> Diana: *"she finds it funny when her brothers make her laugh and make her fall over… she laughs even more then (aww) because she's making them laugh… it's kinda like I dunno.. she's…. Just… she loves her family."*

The intersection between spatiality and intersubjectivity here becomes clear. Home is represented where and with whom she feels most comfortable. This picture of laughter with her brothers presented a sense of comfort and of completeness, where she felt most content and relaxed. A kinship and mutual complementarity is found here in the sense of belonging and the excitement of sharing in closeness. The experience of illness has not entirely invaded this connectedness between her and her family. This resonates with Heidegger's notion of "homecoming", in that we can face up to challenges we are faced with in illness yet come through the other side, and experience homecoming (Dahlberg, Todres, & Galvin, 2009). Similarly to Diana, Glen also expressed Jane's enjoyment of spending time with her family.

> Glen: *"I mean there's always things going I think I think Nick [Jane's brother] keeps her entertained as well really"*
>
> I: *"yeah yeah"*
>
> Glen: *"which is quite quite nice… so there's not a lot of time for her to be to be by herself ... you know be left really [yes] there's always somebody with her or [yeah] you know just watching watching TV with her or giving her cuddles you know"*

This desire for Glen's daughter to be entertained is elicited here. Entertainment is a concept that could potentially speak to the burden created by the illness experience. To be entertained is to be momentarily distracted, to be temporarily diverted from reality. In spite of the value that is presumably attributed to Nick's contribution, there remains a sense that there is little that can be done. There is a dismissive quality to the notion of being 'kept entertained', which suggests that the spectre of illness, whilst temporarily ignored, is never far from his thoughts as a parent. This is not to say that the relationship was not meaningful to Jane, it seemed to lead to genuine enjoyment, but as a family their collective identity and orientation was shaped by the illness experience; in many ways increasing their reliance on each other.

> Glen: *"so yeah she normally finds things quite amusing…but yeah I do sometimes just think she looks so fed up"*

This abrupt return to reality in the extract above suggests feelings of discomfort in embodiment that Glen's daughter struggled with, and the overriding effect this has on the shared experience between parent and child. This follows Heidegger's proposal of homelessness; that illness can cause disharmony or dis-ease (Toombs, 1993). It also echoes Bury's (1982) biographical disruption theorem in the effects illness has on the self, social interactions and daily life. Having people around her brought distraction, a possible description of the 'mobilisation' Bury (1982) proposed, whereby resources are 'mobilised' in strategies to respond to disruption in an attempt to normalise the situation. Yet, this sibling relationship, the intersubjective element was important, meaningful and needed by both parties as a source of comfort. However, in spite of this distraction, there remains the concern that she looks unhappy; this is the sense that dominates the narrative. The illness experience invades even the distraction that goes on around her. Again these moments of change in Jane's mood and in her embodied understanding reveal a sense of not being capable of change, of alienation from her body in the time and space that she is in. Here Glen feels this embodied sense of isolation as he watches his daughter's reactions, monitors her facial expressions and senses her *being* in spite of the relief provided by familial relationships.

Diana and Stephanie portrayed a sense of the importance of friendship for their children. Friends that were able to tell that something was not "quite right" featured in the lives of both, and were especially accommodating in terms of the care and support they offered Elliott and Felicity. Diana explained how Felicity's friends recognised behavioural changes in her daughter.

> Diana: *"…they do realise that something's wrong. I went swimming with them the other day and one of them was saying to me, erm, not in a nasty way but she just said why is Felicity the way she is? And I and I was totally totally surprised... by the question so I didn't really know what to say [yeah I bet] well you know it's just the way she was born she just needs a little bit of help and they they all realise that she's different but they are always helping her…and they try and sort of mother and helping her out and it's it's really heart-warming to see so yeah they definitely know she's different."*

The relationship between Felicity and her friends is shown to be both significant and adaptive to the demands of life with NPC. Friendship in a traditional sense is shown but is distorted by its evolution into a caregiving role as a supplement to normal conceptions of childhood friendship. It can therefore be perceived that even those beyond the immediate family were making adaptations as a result of the illness experience. The idea of disruption or unexpected changes to the sense of self was evidenced in the responses of friends who found their role and sense of self changing to provide a context for the provision of quasi-maternal security in their actions, if not necessarily their words.

> Diana: *"I mean she gets on with things but she's quite forgetful… so in school she's behind on her learning and things like that… so… but everybody in her class keeps an eye on her and everything [yeah] because they've been with her since nursery so … they all know her [yeah] and say it's only Felicity she'll be fine… yeah so they're all really nice and they all help out … you know... they love her to bits so yeah"*

This builds on the notion of a supportive network of peers that know and understand Felicity. It reinforces the idea of the adaptive community whose perceptions of life with illness are shaped by the narrative they encounter. This peer group appears to bring comfort to Diana in the sense of security in understanding but also in providing a degree of normality to an increasingly alien experience.

It must be noted that there is challenge present in the transcript to the narrative Diana conveyed of the understanding peer group. This is hinted at in her response to Felicity's friends' enquiries. She was surprised when one of Felicity's friends approached her with concerns and questions regarding the manifestations of the illness. The shock that Diana felt when this was spoken aloud seemed to confirm what she had been trying to hide, or at least avoid the reality of segregation and increasing separatedness for her child living with illness. This concern about being marked as different, as other, recurs in the next theme when discussing participation in extra-curricular activities. This acts as a useful reminder that the complexity of experience cannot be neatly compartmentalised and that emotions will often transcend attempts to categorise. For Diana, this notion of the fear of segregation can perhaps be seen as a significant concept that unlocks our understanding of her lived experience and identity. It becomes apparent once again when discussing the decision to withhold information about Felicity's diagnosis from friends, a decision that was now being undone by the imposition of symptom progression.

> Diana: *"people in the school didn't know if you know what I mean like the parents and kids and things like that erm ... about a year ago they realised that that… they sort of say is she okay and... they'd see things and say oh I didn't realise cus she looks quite normal [right] and things like that so…"*

The idea of people only recently learning of Felicity's diagnosis seems incongruent with Diana's comments about longstanding friendships beginning in nursery. One would assume relationships have been established yet Diana wanted to keep the diagnosis private. This may have been pursued in order to simply protect Felicity from the potential of rejection or bullying, but it could also be seen as an attempt to normalise life; to live in the kingdom of the well with the other mums and children. NPC was starting to be unveiled and the impact that this would have on other people's perceptions of her daughter, of her, of her family would also be unveiled.

Other caregiving relationships, both formal and informal were discussed. Glen spoke of their experiences with social care workers who would provide care in the home. The staff shortages and high turnover meant that disruptions were regularly seen with regards to consistency of care. Glen made reference to Jane struggling with this and becoming increasingly upset when carers came to their home.

> Glen: *"... she doesn't like new people at all… because she doesn't really accept her (carer)... if she saw her regularly it would be fine but otherwise she's upset... and I can't see her crying [yeah] because we never never make her cry."*

In this extract we can see how a lack of relationship affected Jane. Whilst it is not surprising that the relationship between Jane and Glen was different to that between Jane and the carer, the stark nature of the contrast points towards a significant difference beyond that which would be expected. The distress demonstrated by Jane suggests that the lack of relationship was detrimental to care provision. Glen hinted that he would prefer to have no additional help if this were to remain the effect on Jane, suggesting the gravity of the issue and the significance of relationships. It was telling that Glen was prepared to forgo the assistance of professional help in order to protect both his daughter's wellbeing and his relationship with her. Further to this, it is also revealing that this protective instinct trumped the usual trust in care professionals that is commonly displayed by parents who recognise expertise and cede authority to it. In comparison, Stephanie spoke at length regarding Elliott's carer who attended the home daily. Unlike Glen, Stephanie portrayed a wholehearted trust in Elliott's carer.

> Stephanie: *"…it took such a while [to appoint Elliott's carer] and I don't know if I could go through that again… I think you just get to the point where you're like I might like a night away but erm no I wouldn't want somebody coming in looking after Elliott… unless it's Helen… I'm kind of … reluctant to ask for any help where it might be... different people or… I'd rather just manage."*

> Stephanie: "*I wouldn't want somebody coming in looking at Elliott*"
>
> I: *"No"*
>
> Stephanie: *"I mean Helen has become a member of the family… … we are really fortunate"*

The process it took to build a meaningful relationship with Helen is discussed and we can see the importance of this for both Stephanie and Elliott. The investment in Helen highlighted the trust that Stephanie and the rest of her family had in her, creating a sense of at-homeness through their relationships. This emphasised the importance of care-staff to building rapport and forming attachments with children and the significance that is placed upon gaining trust from the child's perspective. Elliott is described as feeling secure in his relationship with Helen. However, Jane struggled to form an attachment with her carers and this had an effect on the care that Jane would allow her carers to give. The personal care that Helen provided to Elliott merged embodiment, spatiality and intersubjectivity together, welcoming Helen into her/their world. This conjured a sense of togetherness, allowing someone to intimately share your world.

Relationality can therefore be seen as significant for these families as it is the source of both security and insecurity, of diversion and disruption. These relationships provide a context for personal, familial and communal growth, whilst still maintaining an understanding of the severity and seriousness of the situation at hand.

4.2.4 Creating opportunity in the face of adversity

This theme discusses the sense in which the suffering and difficulty experienced by these families served to create the chance for resilience and growth. This was not always a straightforward process, but the narratives which emerged supported this notion. This was seen in that the children demonstrated a recognition of being affected by NPC but found ways to continue to create meaning in spite of its disruption to elements of their lifeworld. This in turn was supported by their parents who wished to keep a degree of *normality* in their lives. Diana longed to do this by pursuing things 'other people's children' do to maintain a level of functional normalcy and to fit in with society; to not allow NPC to deny them experiences that they could otherwise take for granted.

> Diana: *"she does swimming, she's not not a swimmer but she's gaining her confidence in water and she's really enjoying it and she's started doing street dance and she loves dancing and that sort of thing and singing… and taekwondo"*

This desire to share the experiences of her peers and be-in-the-world in a sense akin to these other children, reveals something of Diana's priorities. She can be seen to derive meaning from her daughter being able to take part in the activities in which other children Felicity's age are participating. This helped to sustain Diana's sense of self as a mother, something that is further highlighted in the number of activities that Felicity does. One activity a week may well meet 'ordinary' expectations but participating in multiple activities may suggest an overcompensation against Felicity's incapacity or future incapacity by over-committing to additional activities; to make a public statement of one's ability to cope. It is a stand against the impending temporal threat of NPC. Diana appears to strive for a self-imposed and self-shaped expectation of normalcy, built from experience and perception. The struggle for 'normal' life can be understood as a desire driven by the need for community; an experience that is shared, be it swimming or anything else, is one that resonates with others. It would seem unnecessarily reductionist to attempt to ascribe one particular interpretation and the meanings are probably found in a complex intersection of these competing motives. However, the pursuit of these experiences can be seen as one driven by the expectations of others; the idea that one must maintain the façade, stoically facing up to challenge without yielding appears to be the most significant to Diana's experience.

> Diana: *"we do try to do as much as we can, we go out for days out… she goes to parties she's just been to a girl in her class has erm just had a birthday party where they were rock climbing"*

Diana described situations whereby equality in meaningful relationships was both desired and sought after. Relationships were pursued where they could maintain the same social standing without becoming either a victim or recipient of pity, where the disability would not define them. This was reinforced by the invitation Felicity received to her friend's party. NPC would not be a threat to either Diana's sense of self or the meaning she places on relationships with Felicity's friends and their parents, as well as Felicity herself.

> I: *"yes yes and do you feel like you're missing out on anything or have you had to give up anything as a mum or you know"?*

> Diana: *"ermm no not really no no not really no"*

The repetition of 'no' here could be the emphasis that Diana wants to offer, the sense that nothing has changed, that the family are managing. Yet this stoical approach could represent a means of coping, the drive to keep going, to strive for normality in the broadest meaning of the word; to do more than is expected to show that one's child is well and can take part in these activities, even with the limitations imposed by NPC. Once again for Diana, the fear of being segregated by the alienation enforced by illness drives her to overcompensate in her desire to be perceived as integrated and accepted. It is unclear as to whether this desire is reciprocated in Felicity's own conception of her place within the community, but it is noticeable that her lived experience, relationships and community is shaped by the decisions and priorities of her mother.

As well as adapting to life with NPC from a social perspective, adaptations were noted with regards to changes to the home. An emphasis on the concept of spatiality is shown in how one parent discussed how she chose her new family home with the mind-set that her daughter would need somewhere with a big downstairs space. There was a desire to create an environment that was big enough to manage the effects of NPC, not necessarily in size per se but in a longing for a space that offered peace and enablement despite the effects on physical embodiment.

> Stephanie: *"we chose…well we liked the house because it had a large downstairs space…and it went straight out into the garden as well so it's been lovely really nice garden we can use...lovely downsairs… big wide spaces"*

Stephanie spoke about the advantages of the house that she decided to move to in order to make things easier for her to care for Elliott. However, a deeper sense of meaning is portrayed in that the sentiment goes further than having a physical space to manage the practical implications of caregiving and instead highlights the importance of spatiality. The discussion of "big wide spaces" has connotations of freedom, openness and possibility: an environment where one can relax both inside and outside in the garden. The images that are provoked suggest how spatiality impacts one's sense of embodiment; the physical space provides a steadiness and a consistency, which is juxtaposed with the instability of living with the effects of NPC on the body. The "lovely" environment can aid against the toils of NPC and provide an open space to manage and cope.

> Stephanie: *"he has little foot spas at home… I like to give him like lots of stretching…"*

This picture of Stephanie bathing her son's feet in their home encompasses the united depiction of spatiality and embodiment, giving the optimal level of care, one that fosters relaxation and safety. Therapeutic treatments that can be conducted within the safe and known environment of the home, by a person who is close and meaningful, presented the integration of intersubjective experience through media of space and body. The physical touch, helping him to relax, represents the embodied connection between carer and cared for. It is through the familiarity and proximity of their bodies, the sharing of time and space, that an intimate connectedness and existential concern is revealed – I am here, I will care for you, you do not need to be afraid. This act seeks to relieve discomfort in the objective lived body, the palpable sense of wanting to take away the pain in the present.

Both Stephanie and Glen spoke about their experiences of introducing equipment to their children's lives in order to mitigate the physical effects of NPC. Equipment to aid Elliott and Jane's mobility seemed to not only assist their physical needs but allowed also for a developed sense of embodiment and intersubjectivity. It brought a level of reconciliation to relationships with friends as well as enabling them to take part in activities that they get pleasure from.

> Stephanie: *"…equipment that can... you know... benefit him... we've just got a standing frame for him he has one at school… and it's something that he can be upright in rather than sit being seated"*

This standing frame meant that Elliott was less likely to feel isolated which aided his participation in social interactions with his friends, by limiting physical barriers he would ordinarily have. These physical barriers are inextricably linked to the social interaction at the centre of childhood relationships, particularly where embodied participation and sharing is paramount. This idea of Elliott now being able to be upright rather than slouched showed the opportunity that can be made available through the integration of equipment, the chance to bridge the gap of physical participation as a means of decreasing social separation and alienation. A seemingly simple piece of apparatus can enhance and enrich life in the present. This merges together several dimensions of the lifeworld, bringing emphasis to the wholeness and peace that was sought and created.

A feature of the reflections of all three parents was the adaptation to living with an appreciation of the notion of 'carpe diem'. The mantra of living in the present was a commonality, making the most out of today and not deferring their gratification.

> Diana: *"if we want to go and do things then we go and do it… if it is something we should do with the kids then we do it now."*

There seemed to be a clear recognition of the temporal limitations of life and of not knowing what the future looked like. The future in most cases did not offer hope or security. Stephanie tried to be optimistic regarding the future and hoped that a cure may be found, or at least that Elliott could be enrolled on a clinical trial.

> Stephanie: *"he's lost a lot of his independence I would say over the last 2 years but we're… we're kinda hopeful cus there's clinical trials on the horizon… so we're hopeful that there's going to be some other combination therapy that will kinda just hold him."*

The phrase *"hold him"* is powerful, communicating strong desires for time to stand still as its progress will bring with it further pain and debilitation. This showed a recognition that Stephanie knew Elliott was not going to get better, yet a hope for him to live that bit longer remained. Within this, the idea that Elliott was born at the wrong time was expressed; if only he had been born later, how different his prospects might be. Hurt is felt; science had let Stephanie and Elliott down.

> Stephanie: *"I'm hopeful for other children you know if it's not for us for the other children… there's a lot of hope all round but I suppose it's what's happening day by day."*

Hope was expressed for other children with NPC, that science may be able to help more in the future. This same hope is not shared for Elliott and appeared to be lost to a greater extent when the clinical trials for which he qualified had been set back. Her hope appeared to elicit many expressions, providing energy and drive to continue, as well as compassion for the sake of others. In spite of this, it is this same hope that forces her to confront reality, and this same hope that precedes and exacerbates the pain of disappointment. These varying visions of hope each serve a common purpose, the provision of a goal towards which action can be oriented, and from which a sense of meaning can be derived. Yet even with this in mind, hope's fragility was emphasised.

> Stephanie: *"…there's hope on the horizon and then all of it's dashed because he might not get on a drug trial or… you know everything seems to be up in the air at the moment."*

The uncertainty of what the future brings, in some ways becomes the known to Stephanie. Doubt and insecurity became her expectation. This acceptance that her son probably will not qualify for a drug trial or is too unwell to get better could be interpreted as her allowing herself to admit that hope for the future is fragile. Temporality is key again in this; the future fades into insignificance as forthcoming suffering is known yet its extent remains unknown, the prospect is not welcome.

Each parent's journey through living with NPC holds many commonalities with others: the fact that their child has an incurable disease which affects their present, their future, their body, their home, how they interpret society's perceptions of them, and indeed their own perceptions of how their child should fit into society. In spite of this, adaptations both big and small appeared to create opportunities in a variety of ways. These were experienced beneficially either: physically for the child in terms of overcoming barriers; societally, in terms of integrating the child and allowing a level of normality; or psychologically through an acceptance of the disease and its effects on embodiment and temporality. This was particularly evident in the common attitude of living in the present, where opportunity was more likely to be found than in the future.

> Diana: *"I I I think we tend to not to think too far ahead… we don't know what can happen."*

This theme indicates the changes that children and their parents make in order to survive; what they alter and the ways in which they do so in order to adapt and acclimatise to their new bodies both in the objective sense and in the lived.

4.3 Discussion

This chapter has explored the subjective lived experiences of parents who have children with NPC and how they make sense of how their children experience life with this illness. Adopting a phenomenological approach revealed a detailed account of living with illness. By seeking to understand the children's lifeworlds, the study illuminated the impact the illness experience had on their understandings of the world, particularly embodiment, relationships and its close links to identity. The research illustrated the shared nature of the illness experience and the close connections between the lifeworlds of children and their parents.

Svenaeus (2000a) argued that in the context of illness, the nature of the lifeworld is revealed as one encounters suffering; an idea corroborated by Kleinman (1988) description of the "ontological assault" of the illness experience. Using a lifeworld-led approach enabled a subjective emphasis, the results of which resonated with Heidegger's (1927/1962) "homelessness" concept. Scenes were described where the child with NPC struggled with the objective impact of the body breaking down and how this then transcended to a sense of isolation from the lived world. The body felt alien in that it was the child, but not the child, echoing Svenaeus (2011) description of the body with illness "being me, yet not me" (p.337). These moments described by parents, appeared to impact upon how the child attuned to being-in-the-world, in that the experience of the objective body now felt unhomelike (Svenaues, 2011). In some instances, their mood demonstrated the embodied meanings they ascribed to how they saw themselves in comparison both to the past and to those around them. This experience reinforces the alienation from one's objective body and from the world of shared experience; the embodied relationship to the world had fundamentally shifted. Svenaeus (2011) argued that this is a shift in how one constructs meaning in the lived experience, a disrupted self narrative characterised now by unhomelikeness through the dislocating power of the illness experience.

The momentary nature of these instances of unhomelikeness was illustrated in that parents spoke more of children moving into and living contented lives. The closeness in relationships with siblings and their parents provided a sense of at-homeness for children with NPC especially in dwelling, where belonging was experienced at an ontological level. This co-existence of suffering and well-being emphasised the complex nature of living with illness despite the relative naivety of a child's perspective (Galvin & Todres, 2013). Receiving recognition of these experiences may aid parents and HCPs alike in understanding in more detail the existential trials within the illness experience of children.

The element of temporality was shown to be significant for parents in this study. Heidegger (1962) proposed that humans are future-oriented and that how we foresee the future is based on how we make sense of the now. For some the short-term future would bring death, for others the passing of time would bring deterioration and heightened complexity in the care having to be given. Parents expressed how they had made a conscious decision to try to live in the present, enjoying the moments that they had with their children, complementing other research that has found people living with a LLC and their family carers pre-occupy themselves with the present (Cook et al., 2016; Persson & Sundin, 2008). The relationship of the families to the future was limited by this preoccupation with the present. Death and further deterioration were hinted at throughout the results, but were not dwelt upon; never denied but never outwardly acknowledged either. This points to a larger issue within western culture regarding death. Our failure as a society to reckon with its impending reality has left us ill-equipped to face it (Mannix, 2017; O'Mahony, 2016). Parents and children living with NPC seemed to exhibit a heightened version of this phenomenon, embedding themselves in the present as a result.

Adopting a multiperspectival, lifeworld analysis of illness allowed for an in-depth examination of the experiences of the family as a unit in living with illness. This novel approach was beneficial for several reasons. Firstly, it sought to concentrate on the meaning-making of children living with NPC through the people that know them the best. This was not vocalised through what their children communicate with their mouths but through the language of the body (Kvigne et al., 2002). These expressions of how parents perceive their child to make sense of their illness was looked at through relationality and embodied forms of living together in the intersubjective world. The language of the body of appearing fed up or frustrated at the body's betrayal, shown in the form of mood swings reinforces the body's self-representation (Finlay, 2006; Finlay & Gough, 2003). Movement or appearance as forms of expression should be a component part of research in understanding how children communicate their being-in-the-world when living with progressive illness (Kvigne et al., 2002). The overriding message is that valuable phenomenological data in this study involves the combination of verbal descriptions of the experience of illness and nonverbal language via observations from people who know them the best, thus utilising a multiperspectival approach was a novel and insightful approach to conducting this research.

Focusing on the person-in-context and even the family-unit-in-context has illuminated the importance of holding a family-centred approach for the development of future interventions. In relation to children living with NPC and their parents, a focus on supporting them on the journey of coming to an authentic homecoming is required (Todres & Galvin, 2010). These results have uncovered a need for supporting children living with a LLC and their families through the ontological challenges of living with NPC and the impact that the illness experiences have on their lifeworld. Holistic care at a lifeworld level should be offered in order to meet with some of the existential challenges that have been illuminated through the subjective meanings unveiled in this study. Bergman, Graff, Eriksdotter, Fugl-Meyer and Schuster (2016) suggested that using a lifeworld perspective:

> *"…raises issues beyond interventions, medication and treatments, namely issues concerning our human vulnerability... To have a lifeworld approach to caring is especially important in care settings where questions of meaning and meaninglessness are constantly present."* (p.342).

This is a significant area in which healthcare research should invest in order that HCPs have a deepened understanding of the experiences of how children experience illness at a lifeworld level. The key implication for practice from the results of this study is that it is important for HCPs to have a more in-depth grasp of the effects that living with a LLC can have on the patient and their carers. Further work needs to be conducted looking more specifically at this before recommendations for policy can be suggested. However, the principles of family-centred care should be at the forefront as well as detailed individualised care plans due to the heterogeneity of NPC (Dahlberg et al., 2009; Ham, Berwick, & Dixon, 2016).

4.3.1 Limitations

This study would have benefited from more time being allocated to sustained interaction with and observation of the family unit. As parents felt it was not in their child's interest to be involved in the interview itself, more creative methods could have been utilised to understand the lived experience from the child's perspective. This could have been achieved through conducting an observational study focusing on spending more time with the family unit and what day-to-day life entails. Combining interviews with observation of the families' homes may have provided more depth to the results. Reflective notes from the NPUK conferences that I attended and conversations with children and their families living with NPC were drawn on in my analysis. However, spending time with children in their home environment may have enriched these reflective notes, making it possible to integrate them more into the analysis. It would be beneficial for future research to explore parents' perceptions of this whole programme of work, and particularly this chaper, in order to garner their opinions about my interpretation of the illness experience for children with NPC. This direct involvement would assist in pointing towards a more family-centred and collaborative research approach.

As a method, IPA advocates and encourages an interpretative analysis. Whilst this is welcomed within the method itself, criticism from some audiences may question the validity and reliability due to any differences that may be generated by two different analysts. However, engaging in a ‘thinking dialogue’ within the supervisory team meant that different interpretations of the data were taken into account and an openness to seeing from another’s perspective was encouraged (Shaw, West, Hagger, & Holland, 2016). Due to the prevalence of NPC, it was important that the parents who took part in this study had their identities protected. Although I tried to keep the new characters’ accounts similar to the parents’ original interviews, the process may have affected the authenticity of the parents’ storytelling and how they make sense of how their child understands living with NPC. Adopting this method may have diluted the idiographic component of IPA. However, the subjective experiences of participants in this study were explored in the same level of detail and I would contend that the case-by-case analysis was not compromised.

4.3.2 Conclusion

Living with a LLC has rarely been analysed using a phenomenological, lifeworld-led approach, especially when considering the illness experiences of children. This study adopted this methodology in order to gain in-depth understanding of how children make sense of living with NPC, through their parents’ interpretations. Research would benefit from furthering the exploration of the subjective experiences of those living with NPC in looking more at the body in understanding in a meaningful way the illness experience. As Finlay (2003) argued: “The body discloses the world and in the world the body is disclosed” (p.58). Utilising the above methods could help HCPs gain a greater understanding of patients’ and families’ experiences of illness and could further engender a more humanised approach to care (Galvin et al., 2018).

The next chapter will now move on to explore adults’ experiences of being diagnosed with NPC in adulthood and the implications of the disruptions this brings at a later stage of the life cycle.

Chapter 5 - "I live my life in a desert, it's the only way I can describe it, on my own, walking." Learning to live with NPC in adulthood.

5.0 Introduction

Living with any LLC brings difficulties, but living with a rare disease generates unique challenges for the person diagnosed, their carers and the general public (Joachim & Acorn, 2003). Diseases are defined as rare when they affect less than 5 out of 10,000 individuals (Commission of the European Communities, 2008). Within rare disease groups, the uncommonness of the condition, along with the variation of symptoms, limits knowledge about the condition and how complex the consequences of it are (Jaeger, Röjvik, & Berglund, 2015). This complexity is in part based on the prolonged diagnosis time, the scarcity of support groups and the limitations on appropriate skills and resources that place a particular burden on family caregivers (Jaffe, Zurynski, Beville, & Elliott, 2010; Pelentsov, Laws, & Esterman, 2015). Within the context of this doctoral research, the diagnosis process for NPC can be drawn-out and stressful, taking as long as 5-10 years after initial symptom onset (Burlina, 2014; Mengel et al., 2013; Patterson et al., 2012, 2013). The broad spectrum of the clinical manifestations and the variable age of onset underline the complex nature of the disease and the uncertainty of its related prognosis (Burlina, 2014). Prognosis varies but people who display neurological symptoms in childhood have been shown to deteriorate faster than those who become symptomatic later on in life (Vanier, 2014). This prompts consideration of the disruptive nature of the illness experience for people who become symptomatic during adulthood.

These disruptions are potentially intensified by the argument that the medical, scientific and political communities have to some extent neglected adults with a rare condition through a nominally utilitarian allocation of funding and research; rarity often meaning smaller societal impact than for more prevalent conditions (Angelis, Tordrup, & Kanavos, 2015; Pelentsov et al., 2015). There is a shortage of expertise within rare disease groups due to the under-developed funding structure of work in the field (Garrino et al., 2015; Zurynski, Frith, Leonard, & Elliott, 2008). Public awareness of rare diseases also tends to be poor which could initiate further isolation for the people living with their diagnosis (Remuzzi & Garattini, 2008). Research that has looked at adults living with rare disease has found a sense of discrimination and humiliation in their relationship with and to wider society (Jaeger et al., 2015). This is compounded by a lack of knowledge of the disease that leads to assumptions of the individual's suffering (Jaeger et al., 2015). This is supported by the work of Kvigne et al. (2002) who argued that the societal norms concerning the body and appearance are a source of concern for those with physical disabilities. The awareness that their experience does not correspond with society's demands can therefore lead to embarrassment or shame (Overboe, 1999). This may lead to a sense of seclusion felt by people living with rare disease, impacting on relationships that are already formed or the chance of developing new such connections in the future.

The rhythms and routines of lived experience can be seen as a useful lens for understanding life with and without illness. This idea finds support in the work of Gadamer (1993) who spoke of health as a 'rhythmic phenomena' (cited in Svenaeus, 2000b). A significant change, such as diagnosis with a LLC, has an impact on how an individual engages with the range of actions and interactions that characterise the 'rhythm' of the human condition. Gadamer (1993) couches this phenomenon in Heideggerian terms whereby the notions of being, being-in-the-world and being-with-others are used to demonstrate the relational and societal nature of health and illness (cited in Svenaeus, 2000b). Such terms are statements of our ongoing relationship and connection to the world, the disruption of which through illness and its limitations can lead to a sense of homelessness and disconnection. In a sense, the continual pursuit of the re-establishment of life's rhythm, of equilibrium, can be seen to characterise the adaptation to life with a LLC, yet this pursuit is dependent on others, their reactions and relationships. The individual's sense of being is dependent on the sense of being with others. This rhythmic disruption can also be expressed temporally, in that the relationship between the self and the future is one that is also shaped by illness. The past self, before illness, was able to conceptualise the future in ways that gradually diminish as a result of living with a LLC in a manner similar to that seen in chapter 4.

The homelessness resulting from the rhythmic disruptions of illness can often find expression in feelings of anxiety and existential angst (Heidegger, 1962). If being at-home can be understood as the construction of meaning and familiarity through the relationships and connections that are forged, then the homelessness of the illness experience can be seen as the dissolution of this network. Without this frame of reference, the individual becomes more likely to experience a mood informed by unease and anxiety. Significantly, this kind of angst can also be understood as the springboard for a reconsideration of the relationship of the individual to the world, leading to a more authentic way of living. This is not to underplay the profundity of the disruption of coming to terms with a LLC, but rather it is to acknowledge the complexity and possibility present in adapting to such a change. Through its engagement with these issues of angst and enlightenment, Heideggerian phenomenology can therefore be seen as a useful means of bringing both critique and commentary to living in the world with a LLC.

It is therefore the aim of this study to explore the illness experiences of being diagnosed with a rare LLC in adulthood, with focus on the disruptions caused to rhythm and familiarity of the lived world. This is done to further explore the notion of angst on the lifeworlds of those being impacted by the diagnosis and whether this experience leads to a more authentic way of being-in-the-world.

5.1 Methods

5.1.2 Study Design

An exploratory study design was chosen which was rooted in phenomenology utilising a multiperspectival approach. The phenomenon in this study is living with NPC as experienced by adults and their parent/carer; a carer in this sense being their closet informal caregiver, likely to be a parent or spouse. The study was framed within an explanation-driven logic of inquiry principally using inductive analysis of the data supported by the subsequent use of the existential theories of well-being and suffering to help find the best explanation of the findings (Hiles, 2012; Galvin & Todres, 2013). This study received a favourable opinion from the Research Ethics Committee (ethics number 15/WM/093).

5.1.3 Participants

Five adults who were living with NPC along with their family caregiver - parent (n = 4), spouse (n = 1) - participated in the study. All were UK residents and recruited purposively from the NPUK charity. Due to the cognitive manifestations of NPC in the person diagnosed, two adults with NPC were represented by their parents in the interviews. Interviews were conducted face-to-face in the home of the participant (n= 3), over the telephone (n = 1) or using Skype (n = 1) according to each participant's preference.

5.1.4 Data collection

An advertisement for the study was published on the NPUK Charity's social media pages and interested participants contacted the researcher. Written consent was taken from participants including both the adults with NPC and their family caregivers. The interview schedule comprised of open-ended questions, focusing on the story of diagnosis and the illness experience (see Appendix 9). For adults who did not take part in an interview, as in chapter 4, their family caregivers were asked to put themselves 'in the shoes' of the person they were caring for. Interviews were recorded using a dictaphone.

5.1.5 Analysis

The data were analysed using IPA (Smith, Flowers, & Larkin, 2009) and followed the same iterative procedure as explained in the previous chapter (see 4.1.5 for analysis procedure).

5.1.6 Data handling

Similarly to chapter 4, the merging of participant data was conducted to create three new characters (Hopkins, 1993; Saunders, Kitzinger, & Kitzinger, 2015). This was conducted because of risk to anonymity presented by the small population available for sampling and the strong sense of community present in the population from which the sample could be drawn. These characters were new conduits of the accounts rendered by the participants, attempting to fulfill the dual role of protecting both the identities of those involved and the veracity of their individual experiences. The characters were created by merging together participants who were a similar age and who had lived with NPC for a similar length of time. This allowed for the temporal dimension of the narrative to remain relevant in these composite narratives. In spite of the marginally increased risk to anonymity associated with only merging homogenous accounts, the decision implied here is that the variation of experience was significant enough to warrant a clear distinction in the accounts conveyed. Again amendments were made to information deemed non-essential for this study such as gender, ages and family details, aiming to protect the identity of each participant whilst minimising the potential for significant alteration to the truth of the experiences (Weiss, 1994). Very little in the respondents' quotations was altered, ensuring the authenticity of accounts received. Great care was taken to protect the subjectivity of participants' experiences in order to protect the idiographic commitment of IPA.

5.2 Results

5.2.1 Pen portraits

George was one of the oldest people in the UK to be diagnosed with NPC (see Table 5.1). His wife/carer Tina led the discussion of their narrative concerning the process of being diagnosed and how life had changed living with NPC.

Alisha and her mother/carer, Deborah, spoke mainly about the impact that NPC has had on Alisha's existing social relationships. Alisha lived with her parents and appeared healthy. At the time of the interview, Alisha's NPC remained an unseen condition; she had no physical disabilities.

Josie spoke about her experience of living with NPC. Her father/carer, Ray, made some contributions to the discussion. Josie lived by herself and was motivated to remain fit and healthy.

Table 6. Participant information

Name of participant	Name of carer	Relationship to person with NPC	Age of participant	Age at diagnosis
George	Tina	Spouse	51	47
Alisha	Deborah	Mother	40	25
Josie	Ray	Father	32	25

The themes identified were: *Hiddenness: living within the rarity of NPC; Lonely planet; Loss and diminishing control;* and *Re-understanding and redesigning the self.* The idiographic commitment in IPA meant that a case-by-case approach was taken to analysis. Throughout this iterative process, in each theme, a main protagonist evolved; one participant who features more than others in that particular theme. Focusing on the individuals in this way was chosen in order to present an authentic telling of *each* participant's story and lived experience. This emphasised and brought to the fore the idiographic component of IPA, capturing the subjective experiences in a way that was more authentic and transparent, rather than presenting an 'aggregated' or synthesised set of accounts.

5.2.2 Hiddenness: living within the rarity of NPC

All stories conveyed a convoluted diagnostic process. Initial noticeable changes to adults' bodies varied, but the length of time it took to receive the diagnosis of NPC was relatively similar. Tina felt that she was being effectively silenced after several visits to their local doctor with no escalation of care being considered; these concerns could be 'swept under the carpet':

> Tina: *"you know and over the years he's been to various doctors and I've mentioned about about the eye movements and the coordination and they've said that's alright and I'm thinking well... it isn't ... I don't know anyone else like it... it isn't right"*

This experience of being unheard at worst, and patronised at best, elicited feelings of disregard in Tina and George. This could be seen as a power struggle between the intuition of a parent-carer and the expertise of the HCPs. The picture of the powerful and knowledgeable doctor assumed through socio-historical interpretative repertoires leads to their general veneration and the placing of their judgments beyond reproach; a similar experience to others from within the NPC community as the Foulcaultian clinical gaze is brought to bear on the experiences of different individuals (Lachman, 2013; Wirtz, Cribb, & Barber, 2006). This power dynamic felt by Tina undermined her sense of intuitive confidence as to what was right with regards to health. The recognition that patient intuition could be something that doctors could utilise should be further explored, but it was absent in Tina and George's experience. An equal doctor-patient relationship where mutual-participation is followed was not presented (Mead & Bower, 2000). The repeated visits to the doctor over the same concerns built up a biography that showed help was needed but the feeling that they were unaddressed widened the gap between George, Tina and clinicians.

> Tina: *"… anyways move on a few years and what happened... he was bad… there was deterioration and he was getting worse and he had to go to the A and E…it started from there because they didn't know what was wrong with him… and they done a a referral to a neurologist"*
>
> I: *"mmm"*
>
> Tina: *"… and… and… it was happening"*

A form of vindication was portrayed in the sense of a long-awaited time that Tina knew was coming. She had, in a way, prepared herself already for the night in A&E that had finally arrived. The story had started; the communally recognised deterioration of health was beginning for George. The construction of an illness identity had had already begun to happen from Tina's perspective (Corbin, 2003). Uncertainty as to George's diagnosis occurred for several months, where he was passed on to different specialties.

> Tina: *"so they started delving in looking for things and another neurologist got involved… and they were looking for MS Parkinson's all that stuff motor neurone all that stuff"*

The use of the words ‘delving in’ conjures images of the body as object, flesh and matter, removing the potential impact that this had on George and Tina’s lifeworld. This is further emphasised through George’s case attracting interest and fascination in the medical world, as consultants started to discuss his health due to the almost peculiar and atypical nature of his symptoms. The former disregard of George and Tina’s concerns was no more. The impact of these investigations stirred fear in George and Tina as the sudden seriousness of this became apparent: What was wrong? Why can the professionals not identify and fix this? As George becomes the patient, he ceases to be the person. He was defined by atypicality, by strangeness, alienation and disease. The following journey towards diagnosis was complex and required a number of extensive tests, with NPC finally identified and diagnosed six months later.

The hiddenness of NPC was also experienced in Alisha’s story of diagnosis. Deborah described the consultant’s reaction.

> Deborah: *“... I mean the doctor was really embarrassed because he hadn’t heard of it I mean this was a big professor at a big acclaimed hospital in the liver unit”*

Alisha had been seen by the same consultant for almost fifteen years on a frequent basis; NPC was hidden to the extent that a professor had not found it. The years of unrest and unsettlement whilst waiting for a diagnosis created a sense of life’s narrative being put on hold. Deborah went on to recount how the professor whose care they were under could not face them when the diagnosis was made due to the embarrassment of not being able to give the diagnosis sooner. There is an implicit sense of the challenge to the professional status of the doctor, which in accordance with the power dynamics explored above, may well have been a challenge to their personal worldview and sense of authoritative hierarchy.

> Deborah: *“and when they did that bone marrow biopsy he wouldn’t although we’d been dealing with it for 14 years on a very regular basis he couldn’t face us... and… to be perfectly honest it was only effecting very young children... who weren’t making it beyond 7 so… she was a bit of an unusual [yes] they didn’t know what to do with her ”*

Included here is the impression that Deborah expressed empathy towards that particular consultant and the fact that it took years for him to test for NPC. It emphasised the rarity of living with NPC, and questions how professionals could realistically diagnose the unknown. From a position of hindsight, Deborah could recognise that she was living in a world that did not know the language of NPC. This was further illustrated in Alisha's account of her relationships within her workplace.

> Alisha: *"… I mean the people who work there are physically disabled there's loads of disabled people there"*
>
> I: *"oh really?"*
>
> Alisha: *"lots"*
>
> Deborah: *"you can see what their disability is… this is rather an unusual disease which is difficult… but life is difficult because of the lack of understanding… she looks good"*
>
> Alisha: *"so yeah.. but I mean.. you can actually see them and you can see they are disabled whereas with me... there's some people and they'll never know that I've got got a disability"*

Due to NPC not manifesting itself significantly in Alisha, to the public eye it remained hidden. Even disclosing the diagnosis to her bosses did not solicit much understanding due to her objective body looking 'intact'.

> Deborah: *"and then they think you're drunk sometimes because of your speech"*
>
> Alisha: *"yeah... cus I slur my words as well"*

There is a sense in which Alisha believed it would actually be easier for her if NPC were to manifest itself in a clearer way in order to create a situation where she would be better understood by others. Her colleagues and other people in society assumed that Alisha could do the same as someone who was not dis-abled due to her looking well. This also emphasises the difference between physical and cognitive symptoms in that the general hiddenness of cognitive symptoms creates a barrier to understanding and the few apparent (audible/visible) manifestations, such as the slurring of speech, are more commonly associated with other problems, such as being drunk. These kinds of negative associations were a source of concern and anxiety for Alisha. The societal and communal nature of being is evident here as the lack of understanding and hiddenness combine to create the aforementioned sense of homelessness; a kind of societally enforced exile from the standard modes of support, empathy and common experience.

The problems associated with the hiddenness of the lived experience of life with NPC was developed further in the account of George and Tina's encounters with both the medical establishment and their work colleagues. These interactions for Tina, revealed the tension between different forms of knowledge and understanding that were needed at different times. An objective medicalised knowledge brought clarity in the context of conversations with a HCP but frustration and isolation when with friends from work. When discussing being sent to see a geneticist, Tina said:

> Tina: "*you know I'm so glad I asked and this consultant geneticist when I saw him you know he said… I've been a geneticist for 17 years he said and he said this is the first case in niemann pick… so ask him about it… and he only knows… he only knows the facts as a clinician"*
>
> I: *"yeah"*
>
> Tina: *"and that's the answers I wanted to the questions"*

Tina had been living with a lot of assumptions about George's health for a number of years. This clear approach by the geneticist was helpful to Tina in understanding what NPC actually was as a disease, which had not really been communicated to her before. This extract could be interpreted as Tina preferring to know the facts in a clearer, more objective way, years of uncertainty finding relief in the hands of competence and rationalism. On the other hand, it could be interpreted as Tina only trusting the HCP within the remit of medical knowledge; science being welcome within the confines of the claims it makes. If the HCP had tried to empathise with her or had tried to give her anything other than the facts, would she have listened? Tina saying 'he only knows the facts' suggests a lack of belief as to whether the HCP could relate to their situation and their diagnosis in any other meaningful way. There was perhaps a recognition that the experience and expertise of the HCP could only go so far, that the deep knowledge of what it means to live *with* illness was not the same as knowledge *of* illness. The potential for the parent or carer to feel isolated in their experience therefore began to emerge; nobody knew enough to really empathise with them. This experience of perceived experiential isolation was further shown in how Tina reacted to her work colleagues when they took an interest in George's health.

> Tina: *"… I mean with people at work and... that you see... because you see… and then there's all this information and a couple of the women at work... they say I'll look in the book… what you looking in the book for … you can tell me what it is but you don't understand what you're reading"*

This act of thoughtfulness from her friends showed Tina's belief that trying to understand the biology of NPC was not equitable to the lived experience. Regurgitating knowledge did not show understanding. Now the facts of disease were starting to become less important as the illness experience took hold in the day-to-day of life. Tina had now been told the facts that George was living with a metabolic disorder. Yet that knowledge did not help her or George in everyday life. Here we can see that the body is more than just an object or a physiological organism struggling to function; it is the embodiment of who we are (Merleau-Ponty, 1962). The suffering that was being lived in as NPC started to unveil, and the sense-making of the impact of this on the lifeworlds of both Tina and George, was beginning to be recognised.

Contrary to Tina's experience of her work colleagues seeking to understand more of the disease, Josie found the lack of understanding of NPC from wider society particularly difficult to manage. This stirred feelings of frustration at the lack of awareness and knowledge people have about NPC.

> Josie: *"I'm still learning about this disease but... the way I see it... the more people that understand the disease the easier it'll be for me to live… the easier it'll be for any niemann-pick's patient… for people who don't know about niemann-picks to learn about it…doesn't cost nothing…"*

Here Josie showed recognition of the hiddenness of NPC as she tried to make sense of this diagnosis in her life. Her sense of self-perception was partly informed by a knowledge of NPC that served to explain certain aspects of her being-in-the-world. This knowledge was not present in wider society and there was therefore a sense of dissonance between the perception of the self and the perception of others that disrupted Josie's ability to find a sense of belonging in the world. The perceived lack of knowledge in others resulted in feelings of frustration at the consequent lack of empathy and care that prevented Josie from feeling a sense of at-homeness in her self. This resulted in a lack of attunement with others, preventing her from achieving and living in *Gegnet,* a sense of being in the perfect place to be open to opportunity or to look to the future, while at the same time being happy in the now and experiencing a sense of peacefulness.

This need for widely held medical knowledge of NPC was not found in Tina and George's narrative. For Tina, the perception of *losing* her husband to this disease in terms of his personhood and his character was more meaningful. This loss meant that there would be an alteration to the sense of at one-ness with and belonging to George. The thought of no longer having that sense of belonging with George caused Tina to hide away from him.

> Tina: *"my world burst... ... I was pissed for a couple of weeks I was… I don't know how he felt because I don't recall asking him… cus you don't… you know you know… you automatically assume the worst… what's niemann pick and then... what's in niemann pick and then you think… that's gonna happen tomorrow… right… for 2 weeks… I was like a zombie wasn't I…"*

From her perspective, Tina saw an unjust future of exile through suffering. By her own admission, her initial reaction was selfish in nature, perhaps as she tried to come to terms with the fact that a significant foundation of her lifeworld, her partnership with George, was crumbling beneath her feet. Perhaps such an experience necessitates an inward focus at the expense of others. For Tina, the worst had already happened even though George was not declining rapidly. She was living in the knowledge of what was to come.

Similar interruptions to the lifeworld of the carer were discussed in Deborah and Alisha's story. Deborah spoke about not wanting to worry Alisha with the diagnosis at a young age and so kept much from her. Yet, for Deborah and her husband, the diagnosis being revealed and the resultant consequences caused great fear.

> Deborah: *"I mean it was just So horrible…"*
>
> I: *"yeah"*
>
> Deborah: *"I mean my husband was so... we came out of the hospital and said right we'll drive home and I'm gonna go on the motorway and I'm gonna crash it into the bridge… and I said well I don't think we will do that [mmm] but you sort of drive home and it was just Awful.. cus you sort of looked at this kid and we thought she was fine… and… anyway… that was a long time ago… but no it was awful"*

The revelation that NPC had crashed into their lives seemed too much to bear. Yet Deborah described how she saw her daughter who appeared healthy and could not comprehend that she had a LLC. The label of the diagnosis brought about a change in how Deborah perceived her daughter's embodiment. Although she had not deteriorated when she received the diagnosis, the body was now viewed as being weaker, with-illness, incurable. This resonates with Becker's (1963) writings on labelling theory causing this new sense of embodiment and identity in how Deborah perceived Alisha, even though Alisha was still relatively healthy.

In all three accounts, the misunderstandings or misrepresentations, rooted in a lack of common experience of NPC, highlighted the significance of relationality and connectedness to an individual’s sense of being.

5.2.3 Lonely planet

All three narratives involved a sense of interpersonal loss. For Alisha especially, loneliness was salient. At the start of the interview, before any questions were asked, Alisha commenced with how she found socialising with people difficult to do. As the interview progressed, it became clear that Alisha was making a series of comparisons between her present experience and the friendships she had before her NPC diagnosis. This was evident in the comparison she made between the situations that she and her old friends were now experiencing.

> Alisha: *“I used to have a close friend here who was a really nice guy but he moved to Belfast…”*
>
> I: *“Oh.. oh dear”*
>
> Alisha: *“I mean he’s very very bright... and he used to really help and look after me... [yeah] and we used to go out regularly and stuff and used to go round his house as well and you”*
>
> I: *“yeah”*
>
> Alisha: *“he was just always really nice…”*

The emerging idea of Alisha’s lonely planet comes into view here. The past tense she uses illuminates her perception of the likelihood as to whether it will be possible to speak to this friend again. The disconnection from this friend who has moved away reveals an intersubjective loss that has had ramifications for the temporal sense of the self that she once knew. Reliving happy memories of spending time together, a time of belonging and kinship, shows a level of significance but the fact that this friendship was viewed as a caring relationship questions the extent of the mutuality of the connection. Other past friendships were reminisced about:

> Alisha: *“I think yeah... it’s a problem as well as a lot of my friends have moved away to* jobs *all around the world [I: yeah] and so... yeah”*
>
> I: *“yeah”*

> Alisha: *"and they… my friend Felix... I'd see him almost daily erm but I haven't seen him for over a year so... but he's married now and he's got kids so he's moved on... but I'm still here yeah"*

The reflection that *"I'm still here"* speaks powerfully of the temporal limitations imposed by illness, the inability to move forwards into new experiences, into the traditional markers of maturity and adulthood. A sense of life unshared can be recognised, both in terms of not finding a romantic partner, but also in that the frame of shared meaning and reference between Alisha and friends dissolved as time went on. There was a perception that old friends from school were living a life of which she could only dream, that they were in marriages or relationships characterised by mutual love and affection. The accuracy of these perceptions is almost unimportant, what mattered was the increasing gap between experience and perception of the self and those of the other. There was almost an indication of an unspoken yet agreed upon understanding that her old friends did not have time for her; she thought she could see why when they lived such 'happy lives'. This reinforces the sense that Alisha was being held back by her diagnosis. Her lack of success in terms of career, the stolen chances to work abroad and the inability to form close relationships merged together under the umbrella of living with NPC, creating this lonely planet of isolated experience. The sense of her experiencing 'I can' and 'I am' was not present in her linguistic choices and an ontological insecurity was felt. A sense of homelessness was unveiled here, where the possibility of future kinship seemed non-existent. The sense of embodiment of not having a close, emotional and physical relationship was further unmasked.

> Alisha: *"…as well and I think that's always the thing as well.. I'll.. I won't ever get a boyfriend .. I think that's …I know that's a bit embarrassing kind of thing to say"*
>
> I: *"no it's not"*
>
> Alisha: *"I'll never Ever have a boyfriend … …. …. But erm anyway"*
>
> I: *"yeah … … (sighs)"*
>
> Deborah: *"and there's no answer to it is there"*

This climatic point tied the above utterances of loneliness together with this inner yearning for a close, intimate relationship with another being. One that understood NPC and would not run away from her, but more importantly would understand her innermost being, her embodiment and her identity as someone more than disease. The long pauses in the extract showcased the depth of sadness felt. The heaviness of mood, the sense of being 'closed in' was emphasised through the stark temporal limitations imposed; she had lost her chance. The experience of past friendships, which brought meaning had now been lost to an alienation which would deny the same interpersonal warmth again. The loneliness that Alisha alluded to was also felt by Deborah, who tried to rectify the loss of meaningful relationships.

> Deborah: *"so yeah.. it's an issue the social side of things.. and you... you get exhausted with trying don't you… there isn't anything that fits"*
>
> I: *"yeah"*
>
> Deborah: *"because even people with... niemann pick you know… that's very variable isn't it"*

The presumption that shared experiences of a particular disease could lead to shared feelings and mutually beneficial relationships was alluded to. However, with greater understanding of the disease came an appreciation of the heterogeneity of NPC's symptoms; the fact that no story was the same. The isolation of living with a rare disease was exacerbated by having little common ground with even the other people who are living with this disease.

In contrast to Alisha's despondency at the limitations of a relationship that could occur with another person with NPC, Tina and George used comparison as a way of coping with the loneliness. Many times throughout the interview there was a recognition of George's slower deterioration, especially compared to children diagnosed with NPC.

> Tina: *"…we look at it in the sense that George's deterioration is poor and when I say poor… slow…"*

This could be seen as a way of Tina holding onto the oneness that she felt with George. The normalising of their position reflected a way of coping with this disease; the idea that they had it better than others and that this provided a more optimistic perspective on the situation.

> Tina: *"we're fortunate that his deterioration is slow… you know… but I mean… so you can't turn around and put A time and A date on it… yes I must admit I can see that because when we're at the conference…we have a look around… "*

The element of temporality is key here given that the implicit assumption is that this present situation and this normalising will not last, that George will deteriorate. The mask used when contextualising will be removed and they will no longer be the 'fortunate ones'. This recognition of a limited time horizon aids the present outlook on life, the desire to make the most out of the moments that they have where there is mutuality in their marriage, where they can still share a deep interpersonal connectedness. Having said this, these attempts to place their experience in context were not always present in their account. The long-term dreams of a shared retirement were jettisoned by the illness experience. Tina and George both spoke of wanting to travel and how NPC had robbed them of this possibility.

> Tina: *"this is our time now and we were gonna do all sorts we were right… but… ermm… I'm dubious about going abroad with George now for obvious reasons… we stopped going didn't we…"*

The lack of confidence that Tina now had in being able to go abroad with George emphasises the isolation from their dream that they had planned together. The reduction of spatiality; the world once their oyster, full of freedom and adventure, now closed off and beyond their reach. Dreams were no longer accessible spaces and had been cut back. Their abiding-expanse had withered, and their mobility in pursuit of these horizons had become increasingly restricted; limiting the existential possibilities that were once desired.

Existential isolation was also found in the poignant way Josie described living with NPC, which told a descriptively rich picture of how she understood and made sense of her life.

> Josie: *"… I live my life in a desert… it's the only way I can describe it... being in the desert… on my own … walking…"*

A desert has many connotations of loneliness, barrenness, hunger and thirst. The spatiality element draws upon desolateness and desertedness. The pauses in between Josie's statements emphasised further the sorrow that Josie felt in being so alone. Josie talked about her marriage breakdown and how she had very few friends whom she trusted. Whether this was due to the difficulty of her separation or for other reasons is not known. In one sense, the isolation is felt in terms of her intersubjective self and the lack of relationships she now had, especially the loss of her marriage. However, loneliness was also shown in spatiality in terms of feeling unhappy with where she was living.

> Josie: *"I do struggle to sleep"*
>
> I: *"and is that because of the neighbours or?"*

> Josie: *"yeah I'm have a lot of problems with a guy upstairs... yeah yeah… very noisy…"*

The unrest that Josie felt from her neighbour's actions caused her to feel very unsettled. The home as a place where one can rest and dwell, became a place of unease and discomfort. Loud noises throughout the night appeared to build paranoid thoughts for Josie, noises that were too loud to drown out.

> Josie: *"well… he's ... he's been doing… like… sitting on his bed at night time ... sitting there saying things… so he's trying to get into me head… basically…"*

> Josie: "*he's trying to get into my head and what I'm saying… hang on... if... some… if something's broke… you'll never get into it… my mind's broke… you'll never get into it…"*

The above extract revealed a genuinely considered approach to managing the struggles with her neighbour through the brokenness she felt in her body. This choice to utilise her desert experience in a way that was beneficial to her stood in stark contrast to the pejorative connotations associated with such a barren landscape. Instead it became a place that offered refuge, that offered an abiding expanse where she felt a sense of at-homeness, peace and ontological security. Crucially, it was a place of her own creation; a space defined by her own will, not damaged or disrupted by forces outside of her control. There was an element of Josie regaining a sense of ownership of her sense of self, even if loneliness was the price. The contrast between desert as haven and desert as barren isolation is worthy of note. Even when positively characterised, the function of the desert mentality was still to provide a sense of self-protection, protection from present and future hurt. Yet to make a virtue of isolation is fraught with potential pitfalls. There remains the potential for such isolation to engender a sense of homelessness, a lack of rootedness and connection to the being of others, for which she had previously expressed desire. The desert as a haven could well prove to be an abyss over time.

Nevertheless this transportation to the desert remains an important insight into her ability to cope. She had reached an understanding of *Gegnet,* through which she was offered a place of acceptance of the person she was, of the body she lived in. This became a place to which she could return, a place where she could rest (Pezze, 2006). This was notably something that the other participants had not seemed to have found in their life with NPC.

5.2.4 Loss and diminishing control

The concept of loss was clear and candidly expressed, all were grieving the loss of something as a consequence of living with NPC. For Alisha, loss was shown in her diminishing ability to communicate with others.

> Alisha: *"...somedays I can't find the words I need..."*
>
> Deborah: *"but that's quite a new development... that's only happened this year really..."*
>
> I: *"okay okay"*
>
> Deborah: *"before that you were quite articulate weren't you... that has got worse this year hasn't it... and you can't read very well now either can you"*
>
> Alisha: *"no"*
>
> Deborah: *"... so that's a bit of an issue as well...we try and think of strategies but she's not very good at following recommendations"*

Communication is the very essence of human existence. Not being able to communicate isolates us from the world. The loss of Alisha's voice and the ensuing disconnection from shared lived experience caused her difficulties at work and when trying to communicate with others outside of familiar environments. Every perceived issue could be reduced down to one of impaired communication, which then magnified the loss of Alisha's expression. This had a significant impact on the buoyancy of her intersubjective self, causing a sense of restriction in her social engagements, leading to feelings of entrapment.

Deborah tried to help Alisha remember things through prompt questions, and to some extent did not treat it as a permanent loss, but as something that required work and practice. The message conveyed was that the sense of diminishing control should be fought against. The fight was to be carried out by developing strategies to restore memory, to revive communication, to prevent the future Alisha from becoming more inarticulate and incoherent. As the ability to communicate worsened, the emergent concern was that the loss of intersubjectivity enabled through communication was a threat to the identity Alisha had constructed for herself. Without expression and subsequent recognition, a primary vehicle for the sense of self was fading away. This led to a fear of what the future held, especially with new ‘issues’ that were arising as symptoms of NPC became more apparent and loss of self became greater.

> Deborah*: “but you vary… you have very good days… and very bad days”*
>
> I: *“right okay”*
>
> Alisha: *“yes… yesterday I was so tired I and erm... I I was just stumbling around I fell into a table all... and it was a bad day…”*

Another way in which Alisha’s body was succumbing to NPC was in her inability to control her body. Temporality and embodiment fused together here in how Alisha made sense of life with a terminal disease. A disparity between her old self, that was physically-able, and her present body, that was increasingly less able, led to a sense of resignation and to a markedly different set of expectations and horizons.

> Deborah: *“and it’s the falling over as well”*
>
> I: *“okay”*
>
> Deborah: *“that’s a big problem as well isn’t it”*
>
> Alisha: *“and erm… when I tripped I really hurt myself and erm… yeah… it’s like… but erm… but ermm”*
>
> Deborah: *“you do have some nasty falls though don’t you”*
>
> Alisha: *“yeah I have had some nasty ones…”*
>
> Deborah: *“She can’t correct herself”*

This painted a powerful image of a struggle between Alisha and her body. The self being separate to the body was very poignant for Alisha in a manner that resonated with Cartesian dualism. This notion of not being able to command the body to do the mind's bidding was apparent, almost in terms of being locked into a body that was gradually taking over the decisions that were made. This lack of attunement between body and mind was leading to a growing sensation of gradually becoming unhomelike and uncanny (Carel, 2013; Svenaeus, 2000c). This disconnection also caused the breaking of social codes as to how her body behaved.

> Alisha: *“I was in this restaurant and I walked straight over and hit this table”*

> I: *"did you"*
>
> Deborah: *"and that upsets you doesn't it"*
>
> Alisha: *"it's so embarrassing (laughs)"*
>
> Deborah: *"that's what upsets you because you you... you can't just switch off that…"*

This scene is a manifestation of the impact of NPC on Alisha's identity as a product of her being-in-the-world through perceived social humiliation. Deborah explained how Alisha had ruminated on this experience for a significant amount of time after the incident had happened, a reaction congruent with someone with low self-esteem. Alisha was her own worst critic in analysing the situation, exhibiting elements of paranoia in how she made sense of her identity; what was becoming of her? Why could she no longer control it? A lack of peace was evident throughout this interview as Alisha struggled to understand her body and the diminishing control she had over it.

> Alisha: *"I did this stupid thing and… I know... I had to explain why I did that at the time…"*
>
> I: *did you?*
>
> Alisha: *it must have been hilarious to watch … painful though… yeah I... I said sorry I just didn't see you... I've got a disability"*
>
> Deborah: *"did you Say that to them?"*
>
> Alisha: *"Yeah I had to… I just walked into their table"*
>
> Deborah: *"and that plays on your mind doesn't it... and you keep thinking about that"*
>
> I: *"yeah"*
>
> Alisha: *"yeah… but anyway that's that"*

The intersection between embodiment and intersubjectivity is exemplified here. Alisha felt an otherness to her own body and this then bled into how she perceived others to understand her, resulting in feelings of homelessness as she struggled to explain and justify herself. The pressure to defend her actions emphasised the need for the intersubjective self to still feel part of the world. Saying that she had a disability made an allowance for her actions through which society would be able to understand. This was important to Alisha's sense of identity, especially regarding the sense of 'I am'. She needed people to understand her, to understand who she was in order to bring a sense of security. This mood of shame, frustration and anger was located in the knowledge of the diminishing control she had over her body. Her mood went beyond language and interaction and was a perspective through which her intersubjective and embodied being-in-the-world was understood. An alienation from the body and the lost attunement to the relational world was contained within this account.

For Josie, a determination to stand against her body's betrayal was expressed. Josie had a sibling who also had been diagnosed with NPC during his teenage years. When considering her sibling's situation prior to her own diagnosis, Josie was convinced that she would not develop NPC. She compared herself physically to her sibling and she felt much more able and confident in her 'healthiness'. Josie's father, Ray, started to notice neurological deterioration in Josie and contacted the consultant about this. When the diagnosis came that Josie had NPC, she was perplexed by the results. The mirror representing the mutuality of her future was shown through her sibling and how NPC had manifested itself in their lives. Josie rejected this vision of the future, as evidenced by her emphasis on what she could do compared to her sibling. It was unclear whether Josie was significantly more able than her sibling, as she claimed frequently in the interview, or whether this was more of an act of denial. However, throughout the interview, Josie would perform physical acts to demonstrate her capability.

> Josie: *"cus… well… I can do that (Stands on one leg)… if my brother did that… he'd be on his face… and like... you've just seen… I could have done that all day"*

This seemingly straightforward act of standing on one leg was demonstrated by Josie as a way of proving that she was 'able' and that she could do the things that physically-able people could do. This could be seen as a competitive trait in Josie, a sibling rivalry that could be used to overshadow the dismay she was feeling at her diagnosis. Yet there is also a discourse of battle expressed as she sought to demonstrate the self's mastery over the body. This seemed to be a combination of denial, resilience and defiance in the face of diagnosis.

> Josie: *"you know… then I found out that I had the disease… oh wow… it's nothing to me... I'll show them all at the hospital... I'll show them all all throughout the land... that I'm physically able and I will always be physically able even though"*

Questions arise as to whether Josie fully understood what she was saying, whether she comprehended the changes that NPC had and would have on her life; or whether it was a ferocious spirit that chose not to let her embodied sense of self and identity be ruled by NPC. One acknowledgement which suggested understanding is found in the extract below:

> Josie: *"having lived with …my brother with NPC who was showing signs early on of niemann picks so I've I've been around it all my life… watching you know… like… why can't you do this… and why can't you… you know"*

This understanding of NPC illuminated the rebellion Josie was taking against its manifestations. The illusion of beating NPC, although important to her, appeared to be a way of coping with the diagnosis and a denial of the body's betrayal of her. She was aware of what NPC could do to a person through the loss that she saw in her brother; she did not want to be the same, she wanted to be healthy. Even though there was a strong refusal from Josie to be classified in the same terms as her sibling, there was a soft recognition that she was speaking the words to her body: "why can't *I* do this and why can't *I,* you know." This was evidenced when she spoke about the need for a carer.

> Josie: *"might sound funny… but doing the pots… it really is a struggle"*

Josie preempting what the interviewer might be thinking, attempting to guess the next question, was of particular interest. It showed that she cared how she looked and how she came across. Admitting weakness and allowing me to see this was a significant step, acknowledging the difference between capabilities, explaining her loss and her diminishing control. Priority had previously been given to health, to her physical ability and her resilience. How her body looked was meaningful to Josie as she had spent significant amounts of time "sculpturing" her body in order to make it desirable and appealing. Yet this stage of the interview was the first time that she had really suggested that she struggled. Josie implied other people might find it difficult to understand why she struggled with washing the pots. This might be due to the simplicity of the action and Josie's dismay at losing control.

For George and Tina, loss was expressed in Tina's interpretation of a shifting sense of temporality for her and George's life together, which disturbed their lifeworld and damaged their contentment. As mentioned previously in the theme *lonely planet*, an outlook of living in the present was seen through making the most out of the time they had left. However, this was co-existent with an existential suffering, almost habitual in nature, of speculating what the day was going to hold for her and George.

> Tina: *"…but what's frightening… … … you see… I get out of bed in a morning… … before I get to the bathroom to put the toothbrush in my mouth I do Not know… if I'm standing one step away from a catastrophe…"*

The routine of cleaning one's teeth became the routine of being overwhelmed by what could possibly happen. The juxtaposition of the simplicity of the statement and potential for devastation had a powerful impact. This diminishing of control over even short-term day-to-day life accentuated the overwhelming feeling of concern that permeated their experience. The uncertainty in the prognosis of NPC caused Tina to agonise and fixate. The use of the word 'catastrophe' really emphasised this point; the uncertainty caused tremendous upheaval. As the day began, this tsunami of fear and anxiety began to play in Tina's mind. There appeared to be no rest, no peace. Even in the comfort of their own home, the potential to fall into the abyss was present.

> Tina: *"…because how do I know… that when he wakes up in the morning that he will be able to get out of bed… and walk… or that he'll be able to get out of bed and see… and speak…"*

The lack of control that Tina felt over the deterioration of George would soon begin to rob her husband of his very essence, and with it the foundation of their relationship. This anxiety seemed to permeate through each day and was the overarching response to the illness experience in their narrative. The extent to whether George lived with these anxious thoughts was not conveyed by Tina. However, this narrative stood in contrast to Josie's perspective of an NPC survior and how she conveyed her narrative as within the notion of battle, which was eschewed in favour of inevitable decline and suffering for them as a couple. The fear associated with Tina's daily experiences almost served to provide preparation for the day something does happen. Yet, as each day unfolded, Tina's horizons began to shrink, from paranoia to resignation:

> Tina: *"…. And if you're worried about something you can't do anything about then what's the point…"*

From a carer's perspective, Tina had moments of revelation where she realised that nothing could be done to fix the situation or to stop things from happening to George. Moments like this brought a sense of mobility, a sense of flexible movement forward in experiencing flow and not fearing it. This illustrated the co-existence of well-being and suffering in the temporal fusing with her sense of identity and mood. The fluidity and changeability of living with NPC illuminated the existential confusion provoked by these competing worldviews from a spouse's carer's perspective. There was rest and unrest, peace and conflict, within the overall struggle to find an at-homeness amidst the experience of life with illness.

5.2.5 Re-understanding and redesigning the self

Each participant elicited a sense of re-understanding themselves as a person now living with or impacted by an incurable illness. Josie's chosen manner of living with the manifestations of the disease often seemed to emphasise her physical ability and what her body was capable of doing. Yet there was another perspective that had recently helped Josie to re-understand herself, that of a spiritually-formed worldview.

> Josie: *"... I was going through a big depression that was when I was low... I really low… and i…. does… there's a scripture yeah… I go church… do yourself good... but there's a scripture and… it's Philippians 4 13 … and it says I can do all things through Christ which strengthens me"*

Believing in a higher power, a higher being, provided Josie with a lens through which to view the world in which she lived. It helped her to make sense of her identity, in that it provided a context in which her sense of self was contextualised within a larger, more meaningful framework. This belief invigorated her sense of self in a way that encouraged her to still believe in her capabilities. This frame of reference in which she tried to live her life enabled a sense of mobility, an encouragement of her personhood.

> Josie: *"... the Lord is thy rock... you know… in other words... in my head… it was saying that… it was saying this… as long I focus on Jesus... as long as I keep him in my life… I will have no problems… no problems… because I can do it all through Christ … and I still stand to that today"*

The way Josie offered an understanding of biblical language suggested that this faith she had actively sought and studied had the effect of lifting her mood. There was a sense in which the veneration of the biblical text represented an acceptance of a worldview that moved her sense of self beyond the confines that a medical perspective had imposed upon her. Her desire to overcome had found resonance in a narrative that asserted 'You can' rather than 'You can't'. It was difficult to ascertain the extent to which this re-modelled sense of self had refocused her identity as increasingly spiritual rather than physical as alluded to in her own prior admission of the beginnings of physical decline, specifically in her struggle with the washing up. There are some notable resonances between Josie's potentially shifting perspective and the Cartesian dualism present in Christian thought that might encourage the denial of embodied experience, or at least its subjugation to spiritual reality. More of how Josie re-understood her sense of self was revealed when she stated that she had found inspiration in one of her favourite films.

> Josie: *"well Yoda is a very very wise man and Luke says to him… he says… he was talking about his dad right.. and his dad.. and he… he says. I'll I'll have a fight with him… I'll kill him… no… something … why's it going… come back…. Do or do not… there is no try… he was doing an assault course with Yoda on his back… and he says.. and Yoda's like get up the road get up quick quick… oooo I can't do it I can't do it.. do or do not.. there is not try … so… do it or don't do it"1*

This is a further example of where a presented narrative that claims a particular type of moral or ethical truth is seen to have resonance for Josie. Whilst 'Star Wars' is not claiming the same type of truth as the Christian church, there is a sense in which the totality of the worldview presented created an existential framework in which Josie could make sense of particular aspects of her life. The world of 'the Jedi' is a world where discipline of thought can lead to total control of the body, as well as the temporal and spatial plane of existence. When framed in this manner, the words of Yoda can be seen as powerfully in tune with the worldview Josie had repeatedly espoused, one where the demonstration of physical competence was highly valued. The perceived moral rightness and intrinsic ethical value of both these narratives only served to reinforce Josie's faith in them as legitimate conduits of her own decision making and value shaping.

These lenses through which Josie was making sense of her life, her frames of reference of the self with illness, were not shared by her father within his carer's worldview. Throughout the interview, Josie's father Ray did not contribute much but sometimes provided his interpretation of the day-to-day realities of his perspective of his way of caring for his daughter with NPC. There was a jarring sense of dissonance between the grand narratives that Josie used to provide motivation and meaning, and the fixation on practicality that Ray brought to bear on the experience. The menial task of making a cup of tea was problematic for Josie and Ray's focus on it challenged the utility and realism of Josie's worldview.

> Ray: *"…a very good example and I always use it which is… my generation is… my mum or dad… "put kettle on son"… That doesn't sound like there's any water in… "you didn't… tell me to put any water in dad"*

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[1] "No! Try not. Do, or do not. There is no try." Quotation from the film *The Empire Strikes Back* (1980).

As Josie's world changed her abilities changed also. People in Josie's life such as her father, could see the mistakes that she was making, but Josie could not. This world that Ray and Josie used to share, where shared experiences were common, where understanding of daily tasks was comprehended by both had begun to fade and separate. The distance between her past and present worlds increased, and her father was left behind. Josie had found a sense of spiritual contentment in her desert.

For Tina, a re-understanding of her own identity throughout George's illness experiene was palpable and became an explicit focus when a work colleague came and enquired after George.

> Tina: *"because… …. I'm a carer ... really... I mean… I thought I was that woman that looked after her hubby because and then my manager said that I was a carer… we were having a conversation and at the end of the conversation I turned to walk away and he said you don't do that ... and he said… … ... you deserve a medal you people…. You save the NHS millions people like you… you Carers… …."*

This stark realisation came upon Tina when one of her colleagues called her a carer. She had not thought of herself as a carer until that point. Her identity was still as a wife who helped her husband but something in her sense of self shifted at this point. The hesitation in the extract above portrayed Tina as still trying to understand it, the perception that her identity had changed. She was previously George's wife but now was his carer. How did that look and how did that work?

> Tina: *"you automatically get the job of a carer and I mean… I'm not being funny but when I left work for this… this is the last thing... you know that you know…"*

This lack of preparation for the role, combined with the burden of expectation and the full-time nature of being a family caregiver created a testing experience for Tina. The constant care that George required made life difficult for her. This loss of autonomy made life no longer as care-*free*, instead it became care-*ful*. Tina felt that she did not get much relief from these consistent care requirements that George had. It was a tiresome and weary role that had been imposed on her.

> Tina: *"it is hard to hold your temper sometimes… …. But there's no point in losing it … … because you know You Know… exactly the same thing is gonna happen in 2 hours' or 3 hours' time or the next day… you Know it's gonna be a daily occurrence every time"*

The relentless nature of caregiving meant that frustration became more likely when having to repeat herself whilst helping George with tasks, or when George did not do the things the way that she would have. Tina described tasks performed poorly by George by saying *"it's dangerous for him and it's annoying for me because I'm the one that has to live with it"*. This narrative Tina expressed, seemingly presented a re-education for George, in that he could not perform the tasks that he once could, for example, standing up from a chair. The way he performed this action seemed dangerous in Tina's world and this caused her to feel frustrated that he could not grasp things like he did in the past. This re-learning of place in the world for George was also a re-learning of place in the world for Tina in terms of understanding what George could do and how to manage the situation. Getting used to these changes in George could be difficult for Tina to watch. In some instances there was a sense of bitterness about the life that had been handed to her.

> Tina: *"yeah you see... George has it… has the disease… but as a family… we live with it… that's what people don't understand… … this is my life this is what I didn't want to happen … I didn't plan this... and it's alright to feel like that... it's alright to lose your temper"*

This signified an acceptance of her new role as a carer and an allowance of herself to feel frustrated and grieve the loss of their plans and goals in a way that brought a sense of freedom to feel; a mobility to continue. There were times when Tina felt at full capacity but in acknowledging her position as carer, which seemed to be expressed as living in the *space between* the world of the well and the world of the sick, she gained clarity in the present.

This comparison of the past to what life was like now was also present in Deborah's description, as Alisha's mother-carer.

> Deborah: *"I mean she was in the football team and basketball team and just absolutely... she was get… she went into top form when she started secondary school"*

These clear identity markers of being in the top set, of being physically and cognitively capable, were difficult to shift; Alisha's potential of what she could have been was inescapable. A fact reinforced by Deborah's perception of Alisha also. Throughout the interview, Alisha's opinion of herself was always formed on what she perceived other people thought of her. This was always fairly negative, and often led to the belief that people disliked her.

> Deborah: *"well… you think that nobody likes you that's your problem... any management anybody respon [sic] she thinks they don't like her"*

> Deborah: *"she does have a problem in she always compares herself to everybody else who is... obviously at one stage when she was… Brighter... or able to you know...they all get married they've all got children they've all got in her opinion good jobs in her opinion they all earn tens of thousands"*

A sense of disappointment in her own achievements was felt, this letting her *self* down. This borderline embarrassment of not doing as well as she thought she was going to, led to feelings of self-doubt. Alisha perceived that other people would think that she was a failure too. These issues were tightly focused on the concept of identity as developed through the relationships against which she measured herself. This comparison of the able versus the dis-abled, the opportunities versus the corresponding lack thereof, was how Alisha perceived the world. To combat these feelings, Alisha's Mum, Deborah encouraged Alisha to befriend some of the people that she worked with who have disabilities.

> Deborah: *"because she won't she's she's in self denial.. she saying she's got this stuff and her problems but you won't actually accept the fact that perhaps They're the sort of calibre of people you should be trying to socialise with"*
>
> Alisha: *"mum... I'm always very nice to them at work"*

The word 'calibre' here connotes a particular form of labelling and implied stratification of the social system. The implied notion here was that one is only worthy of mixing with people who are the same as you. This may be one way of Deborah trying to normalise the experience for Alisha, encouraging her to mix with people in *similar* situations, but there was a clear danger here of encouraging a negative and alienated self-concept in Alisha. This was illuminated in the challenge Alisha issued towards her mother's suggestion, which stirred a momentary conflict in the mother-daughter, carer-and-cared-for relationship. Even within the confines of the closest remaining relationships in Alisha's life, there were assumptions and uncertainties about her meaningful relationship to the world as one living with illness. This resonated within the framework of symbolic interactionism, the relationship between self and society. Deborah made sense of Alisha's social networks within the framework of her diagnosis and associated symptoms; she needed to befriend people who were also living with illness. Deborah showed an almost fatalistic mindset in that she sees Alisha first and foremost as ill. This stigmatising of illness into socially constructed groups led to a devaluing of self. This, along with the comparisons made between the past and the present, led Alisha to this sense of negative redefining her *self* as one who is no longer able.

> I: *"Do you get on well with them? Would you Want to hang out with them?"*
>
> Alisha: *"erm no… they are properly disabled as opposed to me whose only got an illness…"*

Alisha spent a lot of time observing others and making comparisons with herself, which reflected a low sense of self-esteem and feelings of depression and anxiety over who she was becoming. This predetermined perspective of her sense of self was reinforced by the perception of 'the well' not wanting to be friends with her, whilst she did not feel 'unwell' enough to relate to those with disabilities. She described a scene when she went out with a friend to a nightclub.

> Alisha: *"and you know and they're all dancing away with each other and there's like... there's just me... and my Poor friend Ian has to probably look after me as well and like…he doesn't have to do that but … it's really nice of him but…"*

According to this worldview, the only way that healthy people could want to spend time with Alisha is in the role of a carer. The use of the term 'poor friend' implies the perception of the self as being a burden to others. It is here that we see the disconnect between the perceptions of Alisha and Deborah respectively. For Alisha, there were differing levels of disability and she was caught between being conscious enough of her own limitations to desire understanding from 'the well', whilst not wanting to be thought of as part of the same category as the "properly disabled." For Deborah, there seemed a more pronounced sense of resignation to the social impact of disability, a more binary view of ability versus disability where Alisha's potential lay in the world of 'the unwell'. Yet where Deborah saw potential and opportunity, Alisha saw a further threat to the sense of self that remained intact and the potential only for further alienation and ostracisation. This conflict and confusion acts as a contributory factor in the pessimistic mood that defined Alisha's illness experience. She was unwilling to face the reality of the redefined self.

5.3 Discussion

This study has explored the subjective accounts of adults with NPC and their family carers. Using an idiographic, multiperspectival, lifeworld-led approach brought to light the existential challenges of living with a LLC. The research illustrated the disruption to the lifeworld that illness had on their identity, especially, when seeking to understand their new sense of self with illness.

The rarity of NPC impacted on all participants especially in the length of time it took to diagnose as a result of its public hiddenness. Budych, Helms and Schultz (2012) explored the medical encounter between patient and physician when diagnosed with a rare disease. They found that insufficient expertise from the physicians meant that the patient would become an expert in the disease themselves, researching the condition and in turn explaining it to the HCPs that they came into contact with. The idea of the expert patient has emerged as a means of empowerment for patients with rare diseases (Ayme, Kole & Groft, 2008). Strategies to increase empowerment within this population includes improving access to medical information and finding support groups in the hope of reducing social exclusion (Ayme, Kole & Groft, 2008). The likelihood of geographical proximity to others living with the same rare disease is low. Therefore online support groups could be seen as more effective in that they allow people to form supportive connections with those in similar positions. From the results of this study, we can see that the heterogeneity of the symptoms of NPC means that it is difficult to share experiences with other people living with the illness. Yet, developing an online community for adults living with NPC may provide a place of support and co-understanding, helping people to re-construct meaningful social relationships and reduce the likelihood of alienation and exclusion.

The disruption that illness caused to the adults with NPC in this study was shown in both the objective bodily alienation and also in how the sense of self was changing. The latter was found to be more prominent and was exacerbated by the sense of the body's betrayal; more developed and longer-lasting cognitive function created awareness of what was being lost. The differing levels of physical deterioration that all participants shared had encroached upon their perception of the self. The effects that living with NPC had on their lifeworld was prominent in terms of the impact on their sense of identity and the changes to their understandings of space and time. The findings of this study resonate with (Carel, 2011) description of the body: "Its dysfunction is so important, so intimately linked to our wellbeing because it is us. Illness is a painful and violent way of revealing the intimately bodily nature of our being." (p.40).

The intimate links temporality has with embodiment were shown in this study. The preference for highlighting the past *self,* which was full of potential and ability, was frequently used as a yardstick for explaining experience. Before illness, the borders of space and time had no limitation; horizons were open. Learning to manage the unpredictability of their bodies was something that all participants discussed. This sense of consciousness of what had been lost is also expressed in the work of Merleau-Ponty, whose metaphorical description of an amputee's experience of a phantom limb is analogous to the adult with NPC who instead lives with a 'phantom future'. Each individual lived with the inescapable memory of the future their bodies were once capable of enacting; they were fully aware of the range of actions and meanings that were no longer available to them. It was a spectre that haunted and defined their temporal and embodied sense of self (Merleau-Ponty 1962, pp.81-82).

A similar consideration of loss has been found in research that looks at other neurodegenerative disorders such as Parkinson's disease (Haahr, Kirkevold, Hall, & Østergaard, 2011), Alzheimer's disease (Todres & Galvin, 2006) and Huntington's disease (Williams, Ayres, Specht, Sparbel, & Klimek, 2009). In Haarhr's study, unpredictability referred to a loss of control and a loss of independence, which was similar to findings in this study. The impression of no longer being able to trust the body was found in people living with Parkinson's disease in a manner congruent with the experiences of people revealed in this study of NPC (Haarh et al., 2010). The taken-for-granted body ceases to be, and in turn alters the sense of space, time and relationships (Carel, 2011).

Research conducted on living with progressive illnesses has shown that people with the disease and their carers take a step back from their relationships (Smith & Shaw, 2016; Todres & Galvin, 2006), renouncing the connections that tied them to the past self. The potential trauma of this is highlighted in the work of Mead (1934) who argued the importance of social interaction in developing and growing in terms of the self and the mind (cited in Aboulafia & Mitchell, 2016). The results of this study served to emphasise the heterogeneity of experience and the varying degrees of meaning individuals placed on such relationships. What was common across all three narratives was a shrinking of the relational world as they sought to understand themselves in the light of illness. This was also accompanied by the desire for meaningful relationships, even if the specifics of what this would entail varied. This need for kinship and belonging highlights the idea that better understanding of the lived experience of adults and carers living with NPC would be beneficial in terms of raising awareness and offering appropriate support.

There are many implications for HCPs working with families with NPC highlighted in the results of this study, particularly in the difficulties associated with variegated illness experiences. This present study found that living with a LLC was a fluid experience, working to live in a body that frequently undermines one's control and independence. These concepts combine to create a complex illness experience, the nuances of which need to be understood by HCPs to enable more targeted care. Carel (2011) has argued this forcefully, seeking to establish an appreciation of the impact of illness on patients' lives as of greater significance than the biological disease. HCPs would, as a result of this perspective, be better prepared to understand that the patient does not reduce their illness to just the physical symptoms, but instead envisions it as a disruption to life, its narrative and their experience. Armed with this understanding, HCPs would be better equipped to support adults and their family carers throughout the highs and lows of coming to an authentic acceptance of their new reality and a feeling of ontological security throughout the challenges of illness (Carel, 2011; Todres & Galvin, 2010).

5.3.1 Limitations

There is arguably an inconsistency of voice across the narratives conveyed where different interviews captured varying degrees of the experiences of either the person with NPC or their family carer. This meant that a degree of flexibility was required in how I responded to the data; discussing predominantly a carer's story in Tina and George's case but more of a diagnosed person's illness story in Josie's case. Although inconsistent, this is helpful in presenting a fuller appreciation of the impact of NPC on the interconnected nature of the familial lifeworld in question.

Occasionally, carers would interrupt when the person with NPC was talking in the interview. It may have been beneficial to have conducted multiple interviews, one with just the person with NPC, one with the family carer and one with both of them. This may have illuminated more of the subjective experiences of both, perhaps showing the extent to which the overlapping, intertwined lifeworlds were fractured by the difference of perspective. However, as an initial basis for understanding the lived experiences of adults with NPC, the current model represented a good foundation for further research.

5.3.2 Conclusion

This study has illustrated that the existential impact of the diagnosis of NPC during adulthood disrupts the entire lifeworld for both the adult and their family carer. Being forced to re-understand their bodies as ill, and to redefine their sense of self in the light of this, co-existed with both suffering in their lived body and an emerging sense of at-homeness. HCPs need to be aware of how the unpredictability of an NPC prognosis and how its toll on the body impacts on everyday life's relationships, horizons, space and movement towards the future.

This is especially pertinent in relation to mobility towards the future, temporality and the sense of identity for both people with NPC and their carers.

The next chapter will complete the exploration of the lifecycle by looking in-depth at the experiences of end-of-life care and the dying experience from two families whose child had died from NPC.

Chapter 6 - Being-towards-death: An exploration of the existential experience of death for two families caring for a dying child with NPC.

6.0 Introduction

In contemporary society, much rhetoric is given to living well and the consideration of death and dying is fought against or denied (Zimmermann & Rodin, 2004). Thinking about death is seen as morbid or negative and dwelling on one's own death may be discouraged due to its melancholic connotations. This has been shown in the way that death has often been described as a taboo (Goldsteen et al., 2006; Kellehear, 1984; Walter, 1991). Historically, there have been varying patterns of responses to the discussion of dying, death and bereavement. Within the earliest human societies, the death of someone would have affected the whole community, which would have been debilitated when its members were lost to death (Kastenbaum, 2013). The Victorian era has been labelled as a 'golden age' for grief, (Littlewood, 1993: p.77) where bereaved relatives would wear black for several months of mourning before moving into a time of half-mourning characterised by the slow addition of "quiet colours" until the bereavement period was over. Philippe Ariés, a French Historian, in his later years researched attitudes towards death in Western Europe over the last Millennium (O'Mahony, 2016). Ariés coined the term 'tame death' in describing pre-industrial Europe; death was not private and there was a familiarity and openness surrounding it. The focus on rituals acted as a guide for people as they were dying or as they mourned.

> *"Death was a serious matter, not to be taken lightly, a dramatic moment in life, grave and formidable, but not so formidable that they were tempted to push it out of sight, run away from it, act as if it did not exist, or falsify its appearances."*
>
> Ariés, 1981 cited in O'Mahony: p.35.

As industrialisation took hold and societies became more urban and secular, the reactions and mindset towards death became increasingly hidden (Walter, 2008). Ariès (1982) suggested that the Enlightenment played a key role in changing society's relationship to death. A progressive worldview incorporating science and medicine undermined the taming of death as traditional community rituals and responses were usurped (Kastenbaum, 2013). The framework in which death was understood was also reestablished as the private nuclear family became the primary means of experiencing social life. A concurrent unease with death led to isolated death becoming the expectation (Ariès, 1982). As a result, the hiddenness of death that governed the later nineteenth century took hold (Kastenbaum, 2013).

These developments can be seen in the grief of the modern West. Death is monopolised by medical institutions and grief seems to be kept behind the walls of the home; these experiences are bereft of community and support, which only adds to the taboo (Gorer, 1965). A humanist worldview took hold as science overtook religion as the primary lens of human understanding (Armstrong, 1987). Kastenbaum (2013) highlighted the medicalisation of death in the modern era as success in preserving life, which has become the measure for judging the medical profession; death is failure. Illich (1976) argued that this is encapsulated within the belief that science would be able to overcome death itself. Death is something to be 'fought', even if that induces greater levels of suffering, opening an ethically grey and difficult debate endorsed by individualism and the fear of accepting the certainty of death.

Heidegger suggested similarly, that human beings avoid mortality, but do this by not thinking about or questioning *dasein* in our understanding of ourselves (Large, 2008; Shariatinia, 2016). We are best understood as beings-towards-death and death is an existential phenomenon, which we must acknowledge and embrace (Heidegger, 1962: 294). Heidegger argued that the 'idle talk of the they' in society, interpret death as one dies but it is not a personalised appreciation of death, it belongs to no one and it is not for the now but for the future. Therefore, we are not expected to face up to death; Death is tranquillised (*Beruhigung*) by veiling its immortality (Heidegger, 1962). Heidegger suggested that this tranquillisation of our being-towards-death affects our sense of dwelling and means that there is a struggle to find a peaceful acceptance of this position as death is viewed as a threat (Shariatinia, 2016; Todres & Galvin, 2006). Heidegger's conclusion of the unique and individual experience of one's own death correlates with an understanding of the unique as an individual experience of one's own life *in the light of death*. In other words, through one's realisation and acceptance of death's non-relationality and its insuperability, one can find an authentic existence viewing death as an 'integrating factor' in order to live authentically, a freedom-towards-death (Heidegger, 1962: p.311). An authentic life can be understood to mean one that is secure and stable ontologically; leading to Heideggerian notions of at-homeness (Galvin & Tores, 2013). Therefore, death, and our orientation towards it, defines our life. In this sense, death can become a means by which we frame our own existence and achieve authentic being-in-the-world (Shariatinia, 2016). The inevitability of our death, our understanding of our mortality, is a vital component in giving meaning to our lives.

Such a recognition of death has to lead to anxiety as we shoulder the burden of responsibility and existence; within the freedom lies the yoke of reality (Heidegger, 1962). Therefore, freedom-towards-death can often co-exist with an existential experience of anxiety and guilt, which calls on our conscience to be aware of the finitude of life (Yalom, 2008). Although these experiences cause distress and are unwanted, Heidegger argued that such experiences play a vital role in allowing us to understand ourselves (Yalom, 2008). Examples of these experiences are the death of a loved one, ageing, retirement; these can awaken us to our everyday being Yalom (2008), by projecting our lives onto the horizon of our death in order to live authentically.

In the face of the modern era, where medicine is highly valued and death is tranquillised, where does acceptance of death come into play when one is diagnosed with a LLC? When one is faced with one's own mortality, does an acceptance of death become more real and in what ways does it cause suffering? How does imminent death affect one's sense of safety and dwelling? Is death fought against when diagnosed with a LLC, even though there is no cure? Paul Kalanithi, a neurosurgeon who encountered death on a daily basis, wrote of his experience of being personally diagnosed with terminal lung cancer:

> *"The lung cancer diagnosis was confirmed. My carefully planned and hard-won future no longer existed. Death, so familiar to me in my work, was now paying a personal visit. Here we were, finally face-to-face, and yet nothing about it seemed recognizable. Standing at the crossroads where I should have been able to see and follow the footprints of the countless patients I had treated over the years, I saw instead only a blank, a harsh, vacant, gleaming white desert, as if a sandstorm had erased all trace of familiarity."*
>
> Kalanithi, 2016: p.121.

Although Kalanithi was well acquainted with death in his treatment of his patients and within the hospital setting, when met with his own finitude, death took on a new meaning, where he engaged with the suffering his diagnosis engendered. Galvin and Todres (2013) suggest well-being and suffering are a continuum and are in relation to one another. They argue that a dual understanding of both well-being and suffering within a care setting can meet individuals in their "vulnerabilities" and their "possible freedom" (Galvin & Todres, 2013: p.98). The dying and death of a loved one is a time where human vulnerability and suffering is experienced. A sense of being-towards-death is exacerbated when faced with a life-limiting diagnosis and the anticipation of death grows as the disease progresses.

Notions of death and suffering are rooted in bodily experience. A fundamental ontological distinction is found between the subjective body and the objective body (Finlay, 2006). The subjective body is the body-that-is-lived, representing an individual's particular view of the world. The objective body in contrast, is the body that is known by the other (Finlay, 2006). "We can peer at, leer at, admire, criticize, probe, investigate and dissect another's body. In doing so, we become aware of it as a contained, material, biological thing." (Finlay, 2006: p.21). This manifests especially in illness, when we can no longer take our bodies for granted. Understanding the objective body could help in understanding the subjective body in a deeper way (Finlay, 2006).

To understand more about the existential experience of dying and death, especially within the modern era, one needs to think creatively of how to do this in a way that is greater than just words, to reflect on a bodily level on one's experience; adopting the concept of embodied interpretation. This approach to furthering our understanding of people's experiences is situated within the idea that the human experience itself is rooted in our physicality, in our body (Finlay, 2006; Hailing & Goldfarb, 1991). Toombs (1993) described how listening well involves close observation of the other's embodied presence. This process involves more than inference; it involves experiencing the whole (Finlay, 2006). It therefore follows that communication between individuals goes beyond language alone, perhaps through what Gendlin (1981) described as the 'felt sense'. Such a sense communicates the experience and interpretation of the individual beyond the words spoken.

Merleau-Ponty suggested that our understanding of bodies is rooted in the bodily understanding of ourselves: "I can understand the function of the living body only by enacting it myself, and insofar as I am a body" (1945 [1962]: p.75). When studying the lifeworld of an individual, their sense of embodiment should be a significant existential dimension that should be taken fully into consideration (Finlay, 2006). Grasping something of the 'Other' both in the objective and subjective is essential within the discussion of dying and death due to the intensity of emotional experience encountered in such conversations (Todres & Galvin, 2006). As Heidegger (1962) states "being-towards-death is essentially anxiety" (p.310) and it is working within this awareness of death that this study seeks to explore the existential experiences of a family's experience of the end-of-life care, death and bereavement of a child to NPC within the framework of suffering. In order to understand in more depth this experience, an embodied interpretation was conducted.

6.1 Method

6.1.2 Study design

This study took a pluralistic approach, rooted in phenomenology, prioritising the lived experiences in order to produce an interpretative idiographic analysis. Further to this, an embodied interpretation was given incorporating the ‘felt sense’. The phenomenon in this study is the dying process (ranging from end-of-life care to after death) of a child with NPC as experienced by their parents. The existential theory of suffering was used to further make sense of the inductive analysis.

6.1.3 Participants

Two families took part in this study who had both been the main caregiver for their child with NPC. The first family consisted of a mother and father whose 9-year-old son had died. They took part in 2 interviews conducted using Skype. The second family’s account was conveyed by a mother whose 4-year-old daughter had died. Similarly, she participated in 2 interviews, one was conducted face-to-face, the second over Skype. Both sets of parents had been bereaved more than 3 months ago, a stipulation of the study in order to allow time for formalities to have taken place. Participants were recruited purposively through the NPUK charity and all were UK residents.

6.1.4 Ethical issues

Ethical approval was granted by Aston University Ethics Committee to conduct this study (project ♯1259). See chapter 3, section 3.8 for details of ethical considerations and procedures).

In the preceding chapters, efforts were made to protect the identities of the participants involved, including creating ‘new’ characters and data cleaning. This is because of the risk of people being identifiable within the NPC community due to the rarity of the disease and widespread community attendance at the annual NPUK conferences. The case study approach adopted for this study meant that gender and age were a rich part of the narrative that if altered would impede upon the idiosyncratic nature of their experiences. I therefore discussed these issues with the participants and gave them options as to how they would like me to present their data. These options were to either use altered details as described above, or to just use pseudonyms, a measure sufficient to meet with ethical guidelines. Both families wished for the central features of their stories to remain in place and therefore preferred pseudonyms. They added that in the unlikely event of them being identified within the NPC community, it would be because of their prior decisions to openly share their stories.

6.1.5 Data collection

An advertisement for the study was published on the NPUK charity's social media pages and interested participants contacted either the researcher or the charity who would then introduce the participant to the researcher via email. Both families took part in 2 interviews; the second being conducted following the identification of further areas of interest during initial analysis. The biographical interview was led by the participants (see Appendix 10 for interview schedule). Interviews were recorded using a dictaphone and data were transcribed verbatim.

6.1.6 Analysis

The interview data were analysed using a hermeneutical phenomenological analytical approach, which was informed by IPA analysis. However, instead of the analysis generating themes, it was written within a chronological narrative of events throughout the death experience. This decision was taken in order to highlight both the sense of flow in the stories as a whole and the overall existential sense-making that each family conveyed throughout their accounts. Like chapters 4 and 5, the existential theory of suffering was used to help better explain the findings from the inductive analysis. Following on from this, an embodied interpretation was practised that engaged with aesthetic phenomenology. The aim of this was to enrich the inductive analysis through engagement in a focused, distinct, embodied interpretation that formed a further analytical layer. The steps below out the analytical method taken.

- I firstly listened to the recordings and transcribed them verbatim.
- I then read and re-read the transcripts, free coding on interesting descriptions, language and concepts related to the phenomenon of the dying experience.
- Having noticed that there was some missing and potentially useful information in the transcripts, I emailed the participants with a number of additional questions. All gave their consent to be interviewed again to provide further details.
- Steps 1-3 were then performed again with the additional data.
- I then made exploratory notes in a Word document based on the free coding in step 2.
- Next, the existential theory of suffering was used to explore in more detail the data and exploratory notes I had made previously. I used each element of the lifeworld to explore how each could aid better understanding of the data, and more specially, the phenomenon in question. For example: How was 'space' meaningful in the dying experience in terms of places, architecture, buildings, landscapes?; How was 'time' meaningful in preparation for death and in the permanency of loss? This offered a lens through which to make sense of these experiences.

- This took the form of an open dialogue between the different explorations that had been applied to the narratives. This involved interrogating the emerging interpretations and sense-making of participants against the elements of the lifeworld and the existential theory of suffering (see 3.6.2 for dwelling-mobility lattice) to identify the most meaningful understandings of the death experience. These findings were written down on large pieces of paper and structured under the narrative timeline.
- The next step was to re-tell each narrative in 4 chronological sections: The first introduced the family telling their diagnosis stories; the second focused on end-of-life care and decision-making; the third explored the dying experience; and the last examined life after death. Re-telling the narratives using these framings enabled a fuller exploration of the events leading up to the child's death, focusing on the existential meanings behind the narrative.
- Using these chronological sections I then re-told the narratives and presented an interpretation of the dying experience.
- Finally, in order to retain the idiographic interpretation of this exploration, the family's narratives were told separately for each chronological section. Similarities between narratives were sometimes discussed but the purpose was an in-depth case study approach.

Following from this analysis, I conducted an embodied interpretation. This was more reliant on a felt-sense than that which was previously conducted. This was presented an aesthetic descriptive piece following each chronological section, drawing more on an embodied reflection. Segregating these embodied interpretations from the initial analysis was done to offer an empathic, aesthetic phenomenological reflection of the death of a child from both families' experiences. Todres and Galvin (2006) who presented their findings similarly in their paper on caring for a partner with Alzheimer's said that they "stand before" the general structures (in this study's case, the chronological narratives of the death experience) as an "embodied being" and imagined, in a bodily and grounded way, what these evoke and signify. Todres and Galvin (2006) argued that it is useful to have one phase of presentation that is descriptive and one that is more interpretive. They argued that it is also useful to distinguish these emphases, which is reflected in the presentation of this study. In this study, both emphases are interpretative, but the chronological narratives focus more on the participants' meaning-making throughout the dying experience and the embodied interpretation includes more of a reflection on my embodied self-awareness; how I as the researcher was affected on a bodily level throughout the interviews and analysis process. I iteratively followed Finlay's (2006) guidelines on attending to the body's disclosure by conducting three distinct layers of embodied interpretation. These layers provide a validated approach, which gave me a level of instruction on the method of this. These are:

- **Bodily Empathy:** This notion goes beyond merely registering non-verbal communication, but instead seeks to understand how recognition of gesture and demeanour in the Other's body imbues meaning and connection between both parties, in this case researcher and participant, leading to a richer and more dynamic listening and interpretation.

- **Embodied self-awareness:** "To take embodiment seriously is to take seriously how one speaks and how one listens to self and other… being a researcher requires that one becomes fully and thoughtfully involved" (Finlay, 2006: p.24). Hailing and Goldfarb (1991) supported this with a similar commitment to reflecting on how the researcher is affected on a bodily level as they listen to and interpret another's experience.

- **Embodied intersubjectivity:** Finaly (2006) suggested "it is hoped that an intersubjective corporeal commonality can enable the possibility of empathy, which, in turn, enables both understandings of the Other and self-understanding" (p.27). This leads to an appreciation that the interrelationship between participant and researcher is a rich and crucial space of communication, empathy and understanding.

I also followed Todres and Galvin's (2008) example of the practice of embodied interpretation. This practice 'carries forward' the meanings that the words and disclosures of the body reveal (see 3.7.2 for more description on embodied interpretation). The method iteratively followed as a result of these considerations were:

- I read through and reflected on notes that I had made during and after conducting the interviews. I focused on the notes that I had made with regards to the participants' bodied presence such as gestures, facial expressions, tone of voice and my relationship to these gestures; how these gestures made me feel and how they helped me to gain a deeper understanding of their experiences.
- I then re-listened to the audio version of the interview, making notes and reflecting on how the participant spoke. This was followed by a focus of attention on how *I* feel, how *my body* reacts.

- I re-read the transcripts and made exploratory notes where I felt something that resonated with me from a bodily perspective. i.e. my heart beating faster or feelings in the pit of my stomach.
- I reflected on points where I felt a connection with my participant, reflecting on the felt-sense of relating to someone, of grasping what they were feeling and mirroring what was being spoken.
- Whilst reflecting on the results of these above practices, I attempted to write my interpretation. This was a carefully crafted process until I felt I had conveyed my felt-sense in the most meaningful way I could.

6.2 Results

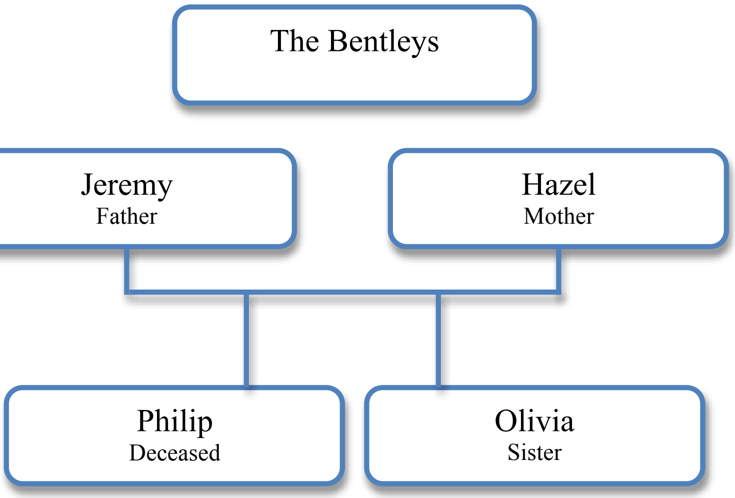
The results below are presented in the stages of the narrative as identified through the analysis. These are: An introduction to both families; Understanding and preparing for end-of-life care; The dying experience; and Life after death. The embodied interpretation follows each chronological section.

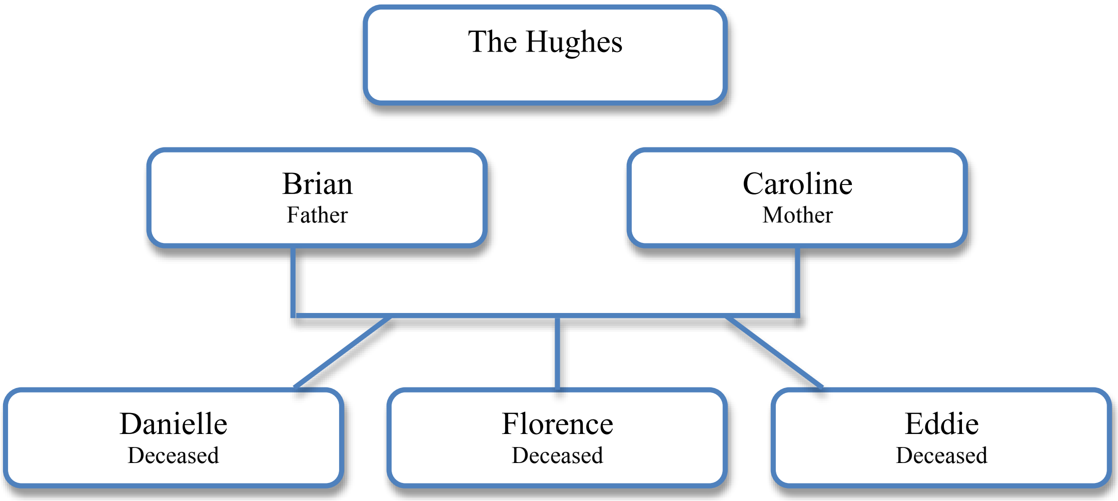
6.2.1 Pen portraits

The Bentley Family: Hazel (mother) and Jeremy (father) shared the story of the death of their 9-year-old son to NPC. Their daughter, Philip's sister, Olivia was present for the first interview. On the whole, Jeremy did the majority of the talking, delivering a detailed timeline of events leading to their son's death. Jeremy still felt the sense of responsibility of caring for Philip, even after his death.

The Hughes Family: Caroline (mother) shared her story of the death of her 4-year-old daughter to NPC. She included in the account some of her husband's, Danielle's father's, experiences at this time, although he did not participate in an interview. All three of Caroline's children had died with NPC, two having been diagnosed gestationally, born prematurely and dying shortly after birth. Danielle's death is the focus of the narrative but all of the children's lives are discussed. Caroline still keeps Danielle's ashes on her bedside table – her most precious possession.

Figure 3. Family tree figures for participant information





6.2.2 An introduction to the families

The Bentley Family

Philip was born after what was described by his parents as a *"traumatic"* birth. Jeremy described there being *"immediate panic in the room"* during the wait for Philip's first breath. Philip looked as though he had stopped receiving nourishment in the womb – as though he had been struggling for months without help. This sense of suffering in the womb was conveyed through the representation of bodily discomfort and pain shown in Philip's body as a baby, something that goes against the expectation of a newborn. The silent nature of Philip's distress was instantly recognised by Jeremy; a visible presence of suffering. Doctors were concerned about Philip's health from birth and at 9 weeks old he had an operation to treat suspected biliary atresia, a rare disease of the liver. A few years later, it became apparent that this was not the correct diagnosis when Philip plateaued developmentally. More investigations commenced and after a skin biopsy, NPC was diagnosed at the age of 4.

The lack of support within community services for children living with LLCs brought difficulty to this family. Their experience was that services existed but in practice were *"extremely difficult"* to access. The weight of responsibility in caring for a child with a rare LLC, where neither parent was medically trained, felt daunting to them. This sense of spatial imprisonment and lack of respite impacted on their sense of freedom as suffering increased. Without support, Jeremy was forced to take early retirement due to Philip's care demands. He described this transition: *"I found that I left one job and moved immediately into another,"* further illustrating the limited freedom during disease and the loneliness exacerbated by community service providers. Overall, Jeremy and Hazel relied on their own skills to research techniques in giving appropriate aid to Philip, as well as applying their own intuition. The only community service that Philip's parents described as being useful was the paediatric nursing service. This was in part due to the level of empathy that the nurses showed when caring for Philip. This is reminiscent of Galvin's (2010) head-heart-hand model of care. In a context where the traditional sources of 'head' and 'hand' knowledge had been found lacking in all but a few cases, it became necessary for them as parents to expand their repertoire of skills. They sought to add both medical knowledge and technical skill to the compassion and love they had for their son.

For several years, Philip suffered many bouts of ill health and decline. His Father described this time as: *"a gradual slow retreat that had new symptoms develop or current ones became more severe… we slowly had to accept this whole difficult situation."* The importance of the temporal dimension is shown here in that it offered the chance for Jeremy and Hazel to grow into the realisation that Philip would not get better. The use of time assisted in their preparation, in their understanding that the future would not appear like the one they had imagined; time allowed for an embodied understanding of the loss to come.

An introduction to the Hughes family

Danielle was born by Caesarean section at 39 weeks gestation. Similarly to Philip, Danielle took around 6 minutes to take her first breath and looked pre-term. She was born with an enlarged liver and spleen, two of the symptoms that are commonly seen in babies with NPC. Like Philip, investigations commenced from birth, and Danielle spent her first weeks in the neonatal unit. HCPs suggested many reasons for the causes of illness that varied hugely in severity; from *"leukaemia"* to *"just wind."* Caroline spoke about how she had previously imagined a more idealistic and romantic life story. One of couples falling in love, getting married and having children.

> Caroline: *"no I just ... I was just completely naïve to the the fact that nothing can go wrong you know especially... all you see in the future you having a lovely family and everything going swimmingly and unfortunately for us it wasn't the case."*

The unexpectedness of complications arising suggested how unimaginable the concept of suffering was for Caroline. A taken-for-granted idea of existence meant that even in the initial complications of Danielle's birth, the preconception that 'all will be well' remained: *"it's just something that happens babies get jaundice you know... she was recovering it will be fine."* This optimism was a part of Caroline's sense of identity, the framework she had lived her life within. As the weeks passed, and Danielle's health did not improve, HCPs began testing for genetic diseases. Although friends attempted to reassure, Caroline did not share their optimism. This dismissal of reassurance suggested the beginnings of a disconnect of the self from others; a felt embodied sense of isolation was growing where Caroline was taking a step back from the life she once knew and stepping into the world of illness. A skin biopsy was conducted which Caroline described as *"traumatic."* She described time as dragging as they waited nine weeks for the results. This trauma was further rooted in Caroline's exchange with a HCP. Whilst awaiting the results, a consultant informed Caroline of his predicted prognosis and the effects that this would have on Danielle's health:

> Caroline [quoting consultant]: *"she would be highly disabled spastic wouldn't be able to do anything... enjoy the memories take her home to die... you've got six months."*

Caroline had no words, she was paralysed in how to react:

> Caroline: *"I had no response to those things that he'd said except to break my heart in front of him…"*

His lack of sympathy for this diagnosis was further evidenced when he asked Caroline, *"why are you crying?"* This shift from the world of well to illness seemed to create a sense of *exile*, a banishment from the life as she had known and imagined; her sense of self seemed to be dissolving. Despite the obvious shock and pain of this memory, its veracity is worth further consideration. Part of why it is so shocking is the fact that it is difficult for one to imagine a HCP avoiding a formal disciplinary after such a statement. This is not to say that the exchange did not happen, but rather that the manner of the account suggests that this memory is being employed for a particular purpose; it is being used to illustrate the hurt that has been carried and the collateral damage to the family's lifeworld across time. Even if this memory reveals only the spirit of what was said, it is still highly revealing of the nature of trauma and suffering. The idea that 'time is the great healer' is made a mockery of in the face of lingering, festering memories that haunt our past. This was not a healed scar; it was an open wound from the past that continued to inform her present and future.

Shortly after receiving Danielle's diagnosis, Caroline was pregnant. This presented a chance to rectify the idealistic narrative she once lived within; to put one foot back into the world that was sickness free. The risk of her baby, Florence, having NPC was 1 in 4, a figure that gave Caroline a sense of hopefulness, adopting a belief that this time *"it will all be fine."* During one of Caroline's scans, it became evident that Florence displayed ascites of the liver, an indication of NPC. She relayed how she *"screamed"* at the geneticist over the phone as he gave her the results of the tests, conveying his belief that Florence had NPC too: the same geneticist who had not long ago explained Danielle's diagnosis to her:

> *"and he just took it he let me scream at him and… and he explained everything and I didn't want to hear it and I didn't want to believe it… it would not go in I just couldn't… you know get over the fact that you know it had happened again…"*.

Her lived experienced was now overwhelmed by astonishment and disbelief. Caroline gave birth to Florence at 23 weeks gestation; a very premature and sick baby. Shortly after birth, Florence died. There were complications throughout the delivery, which meant that Caroline needed to have surgery immediately afterwards. A doctor disclosed to her the news of the required surgery and informed her of potential complications, one of which was that she may not be able to have any more children. Caroline could not cope with this information and told the doctor to leave the room immediately - *"I'm not having it if that's the case right I'm not* *even gonna bother".* She recalled this interaction as insensitive, describing the ease at which the statement flowed from his mouth. This event seemed to have reopened the wound for Caroline, reaffirming her belief at a time of heightened emotion that doctors did not understand her pain. Yet instead of paralysis in the wake of the shock, this time she found her voice, one which challenged the right and authority of the HCP to speak into her world of suffering. With the benefit of time and distance, a listener may conclude that this particular HCP was simply doing their job, but for Caroline it was another part of the growing narrative that defined her motherhood and future. It seemed that Caroline was numb to the risk to her own life. She had lived within the space and time of bodily suffering for less than a year and did not want it to devour all of her dreams of having a healthy family. A few hours passed and she went on to have the procedure, mainly for the sake of Danielle and Brian.

Caroline decided to have one final go at trying for another child. Caroline and Brian looked into additional options that would reduce the risk of having another child with NPC such as pre-implantation genetic diagnosis. However, in the early years of the millennium, this option was not widely available. When Caroline found out she was pregnant with Eddie she was relieved when the 12-week scan went well, with no concerns expressed on Eddie's development; a widening spatiality and a movement into the world she wanted to live in was becoming possible. Perhaps this time it would all be different. At week 16, a scan showed severe ascites of the liver, to which the sonographer, unaware of her narrative, told Caroline "*not to worry… you can have more children."* Caroline reflected on how she felt after hearing these words. She could not move, could not understand:

> Caroline: *"this this is more serious than just 'try again' you're losing a Child… you know you… it doesn't matter how many children that child is still real… yeah and you know the hopes dreams you have for that child are all real as soon as you conceive… and so you know someone saying 'Oh don't worry just try again' it doesn't Work… you know it's not the same…"*

This encapsulates many elements of the lifeworld creating her felt sense of suffering. Scarred again by a HCP's seemingly 'careless' conversation, she was unable to see any good intention from the sonographer. The embodied sense of the pain of carrying a child that was already experiencing disease was serving to confirm her identity, the anticipated yet denied sense of self as a healthy mother or having healthy children. Eddie was born at 33 weeks and did not survive:

> *"it was really really severe and again… incredibly you know just all really traumatic and horrible… and yeah… it took a long time for me to get myself back up you know…"*

The limited words that Caroline could express on this subject emphasised her devastation and was co-existent with the permanent breakdown of the future she envisioned, the world in which she wanted to live, and the essence of her identity.

Embodied interpretation

The brand new experience of becoming a parent initially comes with an acceptance of complications, with uncertainty around what makes a straightforward birth and what constitutes the norm in terms of health when born. The intricate and complex process of a foetus developing in the womb almost warrants the recognition of potential health complications. Yet, the expectancy of being able to produce in reality the idealised picture of a healthy nuclear family is, in the end, assumed. Amidst the whirlwind of the diagnostic odyssey, the uncertainty that HCPs show brings confusion. This seems to incapacitate parents, to prevent them from taking a step back from the situation's fog, in search of clarity. However, a trust in the expert's knowledge leads to a child-like faith in their capabilities. Time seems to stand still as you wait for answers and not just potential explanations. As more complications arise rather than resolve, the more invasive tests start to appear in conversation – time begins to become unstuck as the reality begins to unveil. The seemingly minor aids, such as oxygen given or ultra violet light to help clear jaundice, change to skin biopsies and lumber punctures – procedures that will of course be painful to the one you have just met in the flesh, but already love unconditionally, helpless as they lie down, not able to understand what is happening.

It feels as though HCPs struggle to comprehend the pain you convey, through tears, through anger or through silence. The lack of reassurance offered intensifies the situation. Well-wishers and friends attempt to encourage, to claim a positive outcome through their platitudes. Yet the lack of reassurance offered from the experts, those that know, suggests the bleaker future. Not knowing what they can say, not knowing how to express comfort and instead just walking away to the next plastic cot, to the next distressed family. This becomes an accustomed practice in the place that seems to be a home from home as the faces of professionals become familiar and greet long-staying parents by name. The routine of the ward becomes a flow that makes sense. Confidence builds as medical terms and procedures become understandable; questions can now be asked and understood with regards to inflammatory markers and blood gas results from the numerous heel prick tests each day. Felt tension builds in the pits of stomachs as one sits and observes other parents arrive with car seats to take their babies home; they are well, they are being discharged. An array of emotions are felt; the life that is better, that is ready to start, mixed with the injustice of the life that continues to suffer. The reality of the situation is unmasked as awareness of the severity becomes unveiled; innocence is taken.

6.2.3 Understanding and preparing for end-of-life

The Bentley Family

In the 18 months before Philip died he spent some time in hospital, recovering from a gastrostomy. During this time, Hazel felt that she could not leave his side. Jeremy and Hazel summarised this time as being an "*uncomfortable*" experience as the hospital was not set up for parents to stay with their children. Spatially, the hospital setting was described as being *"peculiar"* which made them feel very *"insecure."* Hazel described not being able to leave Philip by himself; a quick bathroom trip or a rushed lunch in the canteen were the only instances where she felt comfortable enough to leave him out of necessity. The hospital setting prized safety and care as their core ethos but this was not experienced.

Hazel believed the HCPs did not understand that she needed to be with Philip at all times. She had lived within the isolating world of being a full-time carer for several years and knew that other people and HCPs did not know much, if anything, about NPC due its rarity. She therefore showed her distrust in their ability to care properly for Philip. Philip communicated non-verbally and over time his parents knew when he was in pain or the "*very subtle*" physical changes he may show before he would have a severe breathing episode. Jeremy could see that Philip was uncomfortable by *"slight facial changes such as tension around his eyebrows or around his mouth; his body might be tense or his breathing might alter."* This again reflects the 'heart' component of Galvin's (2010) Head-heart-hand model, whereby Hazel and Jeremy, through their relational understandings, recognised the vulnerabilities in their son as a result of the shared meanings of body and world they had cultivated.

Jeremy's sincere claim of an intuitive understanding his son's suffering was rejected by the 'head' knowledge of the HCPs when he informed staff that Philip was suffering from severe acid reflux due to their operating his gastrostomy pump on a fast setting. Jeremy explained how HCP's had *"totally ignored"* his instruction for this pump to be operated on the slowest setting. To prove his understanding, his connection to his son's pain, he resorted to buying litmus paper to evidence this. A sense of *'persecution'* in Jeremy's embodied sense of self was felt in this narrative. His identity as father, as carer, as expert was not mutually acknowledged.

Jeremy used the metaphor of a conveyer belt to explain the hospital setting; where HCPs were given a problem after problem, addressing each in a detached and mechanical way. Jeremy felt that *"for a child that is slowly dying… it's not an appropriate attitude."* This body cannot be fixed. He recalled an instance from his time in the hospital, overhearing some of the nursing team talking about him: *"he's the sort of parent that will tell you that he burnt his bacon this morning."* He interpreted this statement as the staff believing him to be fussy and pedantic. The level to which this comment had offended or affected the way he spoke to the healthcare team was not discussed at first, but it later became apparent that it had put *"enormous strain"* on a situation that was already incredibly stressful. The contrast between the 'conveyor belt' mentality and Jeremy's actions advocating for the care of his son seemed to reveal in Jeremy a clear belief about the essence of a care setting. He had little confidence in a detached and medicalised view of care, and instead continued to promote a person-centred, highly tailored mode of practice, to which he then held to account the HCPs he encountered. This way of thinking also served to highlight Jeremy's understanding of the finality of what was to come, within this worldview there was an acceptance of death.

The Hughes Family

Danielle's condition started to decline in the last 12 months of her life, suffering from regular infections and *"frequent pneumonia."* Caroline described this as the start of the palliative phase where she now viewed Danielle's objective body as beginning to close down. Although there was a level of recognition of this deterioration, Caroline explained how she was on autopilot in terms of providing the best care for her daughter. Routine and structure provided a mobility to care, relying on the daily routine to keep going. For routine to falter would dis-able Caroline, reckoning with the meaning of the reality of the situation.

A peaceful dying experience was something Caroline and Brian wanted for their daughter so they therefore had made the decision to avoid invasive treatments for Danielle once she had reached the point where nothing was going to help; a withholding of and withdrawing from medication that was no longer needed or working. This demonstrated a degree of acceptance of Danielle's imminent death. Yet, when the words were spoken aloud, the reality of the situation dawned.

Plans and preparations are all well and good, and could assist in easing the suffering felt, but the daunting finality of the situation came with the words of the consultant:

> *"I'm afraid that there's nothing more we can do you know… this this… this is the end…"*

Caroline described:

> *"and it's it's hard to hear… even though… even though you've heard it… you know time and again and… she [consultant] didn't hide anything… ever from us… ermm you know we talked about it frequently in clinics… you just think… you just think it's never gonna happen [crying]."*

Acceptance in the present is partial. Knowing the prognosis provided a level of understanding of what the future looked like in terms of Danielle's death. Yet the future was nearer than anticipated or desired. The *'elusive present'* was to be cherished, amidst the suffering and sense of unsettlement, the possibility of inhabiting the present was something that should be fought for, a pressure to make the most of the objective body whilst it was still here.

> Caroline: *"but in the last few weeks it was probably very apparent to other people that she was in the final stages and if I look back at photographs now I can see it myself but as a parent I was just going through it everyday"*

This sense of routine eclipsed the suffering that she had felt previously when losing her two children. Routine covered the bodily deterioration; the chance to really observe, to really fix eyes on her daughter, to connect with the physical presence that represented her only surviving child.

Embodied interpretation

The body breaking down encapsulates the greater levels of suffering felt from both the ill and the loved ones. Being ill is often described as a battle, as something that one must fight. This seems to be especially apparent within the trajectory of particular conditions, including cancer where research charities' straplines draw on the discourse of war, images of beating the disease, of winning the battle. The war against disease does not seem to be immediate, it is instead seen as a series of attempts to buy more time. Attempts are made to ease or reduce the risk of infection by intravenous antibiotics and gastronomies, yet this was not seen as something that would prevent the inevitable. The knowledge that the war against disease and death was long lost came with the diagnosis of terminal illness; it was known that the war effort was in vain. Yet there was still the hope that the final battle could be fought with courage and dignity, that the cause of the living was worth fighting for and that the innocent should be protected. These noble intentions face challenges from questions that arise: How much medical intervention is enough? Why give medication to a dying body? Why fight a losing battle? Allowing the incurable nature to permeate through one's hopes, one's dreams, provides an element of preparation, to foresee the loss that is on the horizon. The sense of loss in all its many forms, of the past, present and future, fading into insignificance as the bodily suffering child is all that can be seen. The piercing words, *"I'm afraid there's nothing more we can do"* is not a surprise, but they violently seal what is known. That which was dwelling in the subconscious now crashes into the conscious.

The realities of the withholding and withdrawing of care begins; the plans to let nature take its course become enacted and embodied. The separation that is the price of death manifests heavily in the body, the weight of it pressing against the chest. Breathing becomes more scattered as the realisation dawns about what the imminent future holds. The felt sense of injustice of 'why now, why us,' presses in. Yet, the knowledge and concept of there being time left, even if it is only days, allows for preparation. The only battle is to shield oneself from the devastation that robs you of the chance to make the most of the time you have left with the person, with the body dying. The hand held is still warm. The shattered heart is put to one side to keep fear at bay. All focus is given to comfort, to walk with and to re-assure, as time draws to an end.

6.2.4 The dying experience

The Bentley Family

Martin House's ethos, the hospice where Philip spent his final days, was tearfully described by Jeremy as a place that "*embraces you,"* that *"put its arms around you and looks after you where you become a part of a family."* Unlike the hospital, there was no felt competitiveness in the priorities, no ambition to fix something that was incurable. Instead there was a calm person-centred focus. A sense of well-being was found in close relationships with staff and other people in the hospice. This served to satisfy the intersubjective self as well as providing an embodied sense of protection and care. The design of Martin House and its architecture was intended to mirror a harbour. Each room faced a mature garden, onto which patio doors would open. The design of the hospice represented *"security,"* which mirrors the notion of the harbour effectively; the refuge in the storm, a place to weigh anchor and find peace. Compared to the imprisonment of the hospital, the multiple floors and closed corridors, the ease and accessibility of Martin's house was described, creating a sense of at-homeness; a sense of beauty and wide expanse within the darkness.

Large windows and patio doors in Philip's room allowed him to look out, to feel the fresh air whilst he lay in his bed; to go outside on his reclining chair and look at the garden, to feel the warmth of the sun. Jeremy recounted a time when he could not sleep so went to get some fresh air. He described the scene, mist over the grass, birds singing, feeling so close to nature, so peaceful: *"I found that unexpectedly very restful"*.

> Jeremy: *"... You know the whole situation was almost impossible... and I remember being on the verge of tears but actually being in the garden with nature did considerably help."*

Finding rest in the midst of death was seen here. This appeared to become his refuge, his calm to settle in the instances where the suffering his son was experiencing became too difficult to manage or witness. An openness in the space offered security and Hazel also commented on the helpfulness of the garden:

> Hazel: *"and for me I was there for 10 weeks and didn't go anywhere else... so had it not been for the garden... if you were in in one place... it doesn't sound that long but 10 weeks to go Nowhere... so the garden it was like... the only place for me to walk round and it was very pleasant... it was good for siblings as well... Olivia learnt to a ride a bike there."*

As well as offering a space to regain composure, it also offered new beginnings; for milestones to be achieved, for laughter to be enjoyed.

Amidst the spacial freedom, the closing down of the objective body led to pain-filled moments as thoughts of 'this was it' were then followed by him pulling through, although incredibly weak. Jeremy sensed that Philip was afraid to 'let go', that he needed reassurance from his loved ones to do so. Jeremy watched Philip one evening as he continually stared at Hazel, refusing to go to sleep. He interpreted this as fear in his son, that he was frightened to die. From that moment of epiphany, Jeremy made a habit of reassuring Philip that they are *"not leaving him, that they will be with him, that there is no need for him to feel worried."* Jeremy believed that this reassurance offered Philip the security he needed to accept death. The self being separate from the body was very poignant for Philip, resonating with Descartes' notion of dualism. When continuing this embodied, mystic relationship that

Jeremy had with his son, he commented on a sense that something had changed in terms of their connection. For Jeremy, this signalled that time was almost up.

This felt sense of the loss of connection could be seen as preparation for imminent death. The pain and discomfort Philip was going through was physically seen, developing a *"huge pressure sore on his ear"* and *"his tummy filled up with fluid… because his system was gradually failing."* Jeremy agonised, as he witnessed the suffering of his son as his body closed down: *"this is Dreadful… absolutely Dreadful."* The suffering was too much. It seemed that he would rather his son be dead and at peace than alive and suffering; death would be a form of relief. After offering the reassuring words spoken above, the apnea alarm monitor triggered. Surrounded by his mother, father and sister, Philip died later that same day.

The Hughes Family

Caroline spoke openly about the bodily suffering Danielle was experiencing:

> Caroline: *"so the morning that week before she passed away… she was pretty poorly and turning blue a lot and having to do a lot of suctioning and oxygen and that kind of thing and I didn't send her to school and I called our clinician."*

Caroline set the scene and spoke about the chaotic nature of their family home, which was *"full of community nurses."* She described how *"our clinician pretty much lived with us"* with value being placed on the word *"our."* It was not a stranger involved in the end-of-life care for Danielle, but someone who they had been on a significant journey with throughout Danielle's illness. They were being supported by professionals who felt like family, a stark contrast to the care received when Danielle was diagnosed. A change in relationality was seen here. There had been a shift from feelings of being marginalised and misunderstood to feelings of kinship and community. HCPs were no the longer the enemy, they were an ally. This was perhaps down to the imminence of death serving to fundamentally shift Caroline's perspective; the uncertainty and disappointment that framed the previous encounters with HCPs was now gone, replaced by a certainty of outcome that enabled being-towards-death.

Caroline spoke with a sense of nonchalance when describing the many allergies that *"their"* clinician had and how they had to *"completely change our cooking"* due to her being a vegetarian: *"it was completely mad."* These finer details were typical of the clarity with which Caroline remembered the last 7 days of Danielle's life. These observations also served as a deflection away from more painful moments, the busyness of life rather than the suffering of her daughter. Caroline described how her husband found the chaos of their home difficult to manage. Brian had not received the sense of closure that Caroline had as a result of him not attending the hospital appointments. For Brian, his home had seemingly become artificial, an unknown environment. His own feeling of being unprepared for what was to come was magnified now through his home becoming 'a hospice'. It was too challenging and he would often escape to the outside, air was needed.

In a similar manner to that described above, Caroline then went on to speak about the evening that Danielle died. Times were given as she described the scene as if it happened yesterday.

> Caroline: *"Around six o'clock in the evening ... he [Brian] bathed her and so she was all lovely and her hair all plaited and everything in her nice favourite pyjamas."*

Feelings of comfort, contentment and relaxation are captured in the quote above. Time with just Danielle was something that Brian had needed as he prepared himself for her death. The intimate act of bathing his dying daughter, holding her in his arms as she relaxed in the water offered a realisation of what was imminent. This allowed a chance to stare at his creation, that which he had reared, gazing upon her flesh, reminiscent of the day she was born. Caroline described how she then wanted to follow suit and take a bubble bath. The tumultuous nature of the preceding 6 days had taken its toll: *"so I went and had the most amazing bubble bath relaxing bubble bath an came out feeling wonderful."* This sense of relaxation numbed the suffering. Repeating Danielle's action seemed to embody a shared experience, crystallising a memory of what her daughter had just taken part in, providing an experience that she would not forget. Even many years later, this bath was still described as *"the most amazing"* experience. This connection, to the flesh, of her flesh, would be something that she would always share with Danielle.

Serenity reigned in the moments that followed; calm after the storm of the last 6 days. For the first time in a week, the community nurses and their clinician had gone home. Caroline continued the description of the evening, the time mapped out her actions. That evening created a sense reminiscent of times past, all three together. They decided to order a Chinese takeaway, a further sign of feeling the familiarity of their own home. A sense of them being able to breathe again and enjoy their evening together.

In the midst of her peaceful home, with her parents by her side, *"...Danielle decided it was time to go."* This was not conveyed as a shock but as expected, even natural. Perhaps I could have asked more about this, but the sense of peace that Caroline conveyed was the essence of her memories. Danielle died in peace.

> Caroline: *"Danielle was comfortable the whole time she was not in pain at all… the pain relief she had done was all done amazingly well…"*

In those moments, suffering did not seem to be experienced. Caroline half-smiled at the idea that Danielle had been waiting for it to be just the three of them before she died, a moment of levity betraying her doubt as to whether this was really the case scientifically. This was revealed further when she said:

> Caroline: *"…and you know the final thing she did was smile… and you know whether that was an involuntary reaction which some clinicians would probably tell us I don't care… … [crying] I don't care… I think she was because she was just waiting for us and you do hear it…"*

The need to add this disclaimer may be a reflection of the harsh world of medicine that Caroline had encountered throughout Danielle's illness journey. The insensitivity shown by HCPs when talking about her daughter's prognosis may have clouded why she chose to rationalise her daughter's peaceful death. Was her daughter's final act an expression of love or an *"involuntary reaction"*? The idealistic world that Caroline had lived in was hinted upon again here, even in such a moment of pain and suffering. The narrative she wanted to believe in was being challenged again by the unrelenting rationalism of the medical community. Would that smile, like the family she had wished for, be robbed from her by unsympathetic, uncaring biology? Caroline carried forward the wounds of past hurt, even as she clung to a vision with which she had made peace.

Yet, the overriding picture presented was the finality of Danielle's earthly narrative wrapped in a loving moment shared with those she loved most, a moment from which Caroline drew comfort. Caroline telephoned their clinician, who came back to their home to do the formal checks and requirements. She then left and Caroline, just as she had done with Florence and Eddie before, was able to spend the night with Danielle:

> Caroline: *"and we had a lovely peaceful night… just the three of us together…"*

Embodied interpretation

The intimate moments shared before death comes, depict its strange beauty. The preparation for a peaceful rest is met with favourite pyjamas and hair plaits, adding beauty to the coming darkness. The familiarity of preparing a child for bed, readying them for sleep, is met with the knowledge that this time there will be no waking. For the child, there are feelings of security and at-homeness as the nearest and dearest surround. Calm envelopes the experience, shown in a smile as the last breath is breathed. Suffering had finished for the child, replaced by eternal rest. The surroundings are familiar but now the flesh is free once again from machinery, from the sounds of beeping equipment and strangers coming to monitor the body in its ending. The sedated body still seems to feel, the comforting hands and words of loved ones gently communicating the depth of relationship as the body fades and stops.

Yet for others, the experience does not seem so serene, death seems to linger at the door, wavering in its decision as to whether today is the day. The body somehow unhooked from its grasp to live another day; uncertain as to when it would finally be allowed to rest. This hesitation prolongs the moment of suffering for both the ill and the loved ones as the working body painstakingly becomes alien. Observing the pain and the suffering, watching the body closing down, fluid building as the body is no longer able to control itself. Amidst the darkness of suffering, the felt sense of peace is available and present. It is seen in the spatial appeal of mature gardens shown through big windows as the light shines through and is entwined with the presence and whispers of loved ones that comfort the dying body as it slips away. For a brief moment, time seems to stand still as an embodied relief is felt; pain has finished and will not be felt again. This relief is soon met by the overwhelming sense of loss that takes up permanent residence, as the life known and loved ends.

6.2.5 Life after death

The Hughes Family

The morning after Danielle's death, the funeral directors arrived to *take* her body. Caroline described them as being *"wonderful"* and *"really nice the way they did everything."* The lack of detail given in this section may be due to the pain that was felt when Danielle was taken away from her home for the last time. The knowledge that she would not return, that she would not sleep in her own bed again or be present in the home may have been too much for Caroline to talk about. There was a sense that the perhaps the memories, hopes and fears that Danielle's embodied presence represented were under threat; the absence of the body meant the loss of more than just that. Yet there was also contentment at the manner in which this embodied manifestation of her love was treated with dignity and respect by those that took it away.

In the week leading up to the funeral, Caroline described herself again as being on autopilot during the legal procedures, plans and visitors that Caroline and Brian had to manage. Although pandemonium was in the house, when asked again how the house felt, Caroline juxtaposed this feeling with emptiness. The house, the epicentre of family space, was the busiest it had been, yet felt vacant. This feeling was very present when two days after Danielle's death, social services came to collect the equipment that had meant Danielle could live at home.

> Caroline: *"I know they're not her things because I know you know… they're lent to us for that purpose… but they are a part of the family… that's her bed that she slept in… chair that she sat in you know… her wheelchair or buggy whatever you want to call it and her bath chair… all of those things…"*

This removal of Danielle's existence was too much for Caroline to watch. She shut herself in a room and left them to it. The bath seat, which Brian had used to share in his favourite time with Danielle had been taken away. A lack of recognition for the life lost was shown by social services. Caroline knew that this would be inevitable, that the social services would need this equipment back, but the day after she had died seemed too soon. The bed in which she died had so much meaning, the space that had held and enabled the embodied self was being taken away. It seemed that the peace of the ideal death had left and the emptiness of reality had replaced it. A heavy gloom characterised the mood of the home; the awareness of past wounds reemerged.

To get away from this, Caroline along with her husband visited the coastline on a daily basis in the weeks after Danielle's death, living on coffee and bacon sandwiches from a beach hut: *"and that was all we could… just let the sea air blow everything away."* Home had once again become a place of *roomlessness*. Being outside released this sense of entrapment and offered them a chance to escape as they allowed the wind to sweep over them and began to breathe again, preparing for life as a family of two.

An instinctive almost pre-prepared acknowledgement of what the week leading to the funeral would be like had been dwelt upon before Danielle died. Caroline explained how she was not the type of person who would *"collapse like in a heap on the floor"* and instead focused on the job in hand, reminiscent of the routine she had identified herself with when Danielle was dying. This way of managing the situation was not handled similarly by other family members, which appeared to be a frustration for Caroline.

> Caroline: *"… as parents it's us that perhaps should be the ones who were being hysterical and unreasonable and you know having a melt down… Not when you're telling family members you don't want them to be hysterical and having a melt down ermm because really… it's not about you and I I get it… it's upsetting but they can go away and be upset and not show it"*

This level of empathy shown by others regarding their situation was something that Caroline did not perceive as empathy, but rather as an invasion into her space and identity as a mother by expressing grief in such an open and 'indulgent' way.

> Caroline: *"…sometimes I was calling people and sometimes they wouldn't be able to speak on the phone and I can understand that but that's not what we need… we just need you to be calm as we are…".*

A sense of isolation had been lived in for sometime and this would continue. Empathy did not help; others finding the loss of Danielle very difficult was not a source of comfort to Caroline; she was imprisoned in her own narrative, where no one else could be helpful. They had not lived through it, they could not understand. Finding a sense of calm amidst the chaos was something that was shown throughout both the week leading up to Danielle's death and the week leading up to her funeral, with her husband who shared in understanding and experience providing the space that she needed. Her nuanced and unique lived experience was here writ large.

Further thoughts were given to how other people interacted with Caroline and Brian after Danielle's death. People would often try to find the silver lining, or encouraged her to 'count her blessings', a reaction that Caroline found increasingly frustrating. There seemed to be a conscious decision that it would be better to continue to alienate themselves from others. It might be more a case of no one could say the 'right' thing. The kingdom of the sick, in which they had been living, had separated them from others in the kingdom of the well, unable to cross the border. Conversations with others who had experienced NPC were received, hinting at the significance of shared experience in losing a child and the depth of relationality felt.

This continuation of living in the kingdom of illness was shown again later in the interview when Caroline described how she and Brian had moved house since Danielle's death, but that Danielle still had her own bedroom in their new home. There was here an appreciation of the connection between space, memory and relationality; their embodied space needed to tell their story.

> Caroline: *"it's it's a little girl's bedroom and she's not a little girl anymore… you know she'll be 14 this year… 15 this year 15 in May… so it's about time I redecorated".*
>
> *Caroline said that "people may judge us but we have to accept that's part of it."*

Caroline appreciated that other people may not be able to accept these decisions. The sense of maintaining connection with the dead, of still wanting to be in the world with her daughter was present here. This was one reason why isolation was preferable. She did not need others to gratify or condemn her path and how she had chosen to walk it, it was for her and her only.

In the months that followed the funeral, Caroline described how she would wake in the morning and forget for a moment what had happened:

> Caroline: *"…you know you go to sleep if you can go to sleep and you wake up the next morning and you think 'why hasn't the monitor gone off?... oh yeah that's why the monitor hasn't gone off' and you know you re-live it all again and so for me I think the mornings were always the worst… ermm because you are you are just thinking again and Before you go to bed because it's quiet and you've got nothing to do and you do go to sleep and wake up and you've forgotten… just for that instant minute"*

> Caroline: *"…did we do the right things did we follow the right steps did we choose the right care… I know in my heart that we did… but I still question that myself you know… did I sponge her mouth enough did I do this… you know were her feet comfortable you know just all these different things come to you in the middle of the night and torture you and you just want to know that you got it right…"*

In the dark of night, reality seemed to be corrupted as confusion and doubt inhabited the mind. Thoughts persecuted Caroline as she struggled to rest and instead dwelt on Danielle's suffering and contended with her own. The felt responsibility weighed heavily on her. These thoughts could not be answered but she knew in the light of day that she was now the one suffering. Her identity as a mother and carer being tormented, as they had been so many times before, this time the change to make amends would not present itself. As time progressed, the sub-conscious acknowledgement of their home now being a quieter place took hold, and the reality of the situation sunk in; the flashbacks became fewer in the mornings, as did the sleepless nights, only recurring around the times of the anniversary of the children's deaths. When asked about a plaque or gravestone for her three children, Caroline revealed that Danielle is always with her:

> Caroline: *"… I've got Danielle's ashes with me on my bedside cabinet and they're in a lovely pink wooden box with pink gerberas on all over so those are the flowers we chose for her… so it sits on my bedside cabinet and it's got peppa pig on the top because that was her favourite… so she's always there… I always talk to her and say good morning and good night…".*

The need for her daughter to be next to her illuminated the need to be present with her daughter in every way possible. The box was described as being the most precious item in her house. The remainder of Danielle, in dust, was something that Caroline wanted to have right by her as she slept, in the intimacy of her bedroom. Shut away from any communal space, there she could be with Danielle. For Florence and Eddie: *"I've got footprints and hair and I have all of those things… I can't look at… but they're all locked up very tight in a drawer."* The knowledge that she had those special paintings brought comfort to Caroline. These pictures cannot be unlocked, as this would be too painful for her. Physically holding the paper and looking at the footprints or hair would make things too real in the present and would invade and depress her mood. Caroline spoke of how she had thrown herself into work as a way of moving forward, of still being able to live. Yet around the time of anniversaries, she came to a standstill as she relived the memories of her children's deaths, and imagined what they would be doing now if they were alive. Imagining the family that she had envisioned before she fell pregnant with Danielle.

Sometimes on these anniversaries, the drawers were unlocked and photographs were looked at and she allowed herself to be enveloped by the pain she normally pushed off through routine. Yet, to keep moving, she locks *herself* away again, with her most precious of possessions that are *"locked up very tight."* It seemed that Caroline could either move or dwell, it is not yet possible for her to *peacefully* do both at the same time. Dwelling awakened the painful memories, movement allowed her to keep living in a way that did not cause her to be unstable. But outside, in her garden, in the fresh air, where she can breathe, three memory roses for each of her children grow.

The Bentley Family

Jeremy described the moments directly following Philip's death as he stayed and helped the nurse wash Philip down and prepare him; a process in which they took their time, showing gentleness and making the most of those moments, not wanting to rush away. Philip was moved to the cool room; a room designed for and belonging to death. He was wheeled on a trolley into this room where he was then placed in a bed. The family went onto say how they stayed at Martin House up until the day before the funeral. When asked why they had decided to make this decision, their reasoning was simple *"in order to be close to Philip".*

> Jeremy: *"If we needed… to… go have a word with him or share our thoughts… then we could go down [to the cool room]… so Philip was there"*
>
> Hazel: *"and it was just a normal bed wasn't it"*
>
> Jeremy: *"yeah"*
>
> Hazel: *"in a normal bed"*
>
> Jeremy: *"so… he's there and with you… then he passes away but physically he's still there"*

This was understood by Philip's family as a helpful, and in many ways precious, transition from life to death. The body being there to see and touch was significant to Jeremy and he sympathised with parents whose children had died whilst they were in hospital or at home; *"they miss out on being able spend time with their child"* in these settings due to the legality or cultural decisions made when the body is taken away. The loss of the body, wrapped in history and memory, was symbolic of the loss of self. Hazel said another reason for staying at Martin House was because she had not been back to their home whilst Philip had been in the hospice, 10 weeks in total. This felt like a significant amount of time and had left a distance between how Hazel especially felt about the home. When they did go home, she remembered it vividly:

> Hazel: *"washing that I'd done and I'd hung some of the things on hangers were still hanging up that was really surreal… that's one thing I remember and I said to my mum 'why is this washing still there? ... it was just strange that I hadn't been home for all that time so that was… some things were still the same… it was really strange coming back after all that time…"*

During those 10 weeks spent at the hospice, a sense of time being put on hold was felt; a world frozen between life and death. There was a reluctance to leave the hospice and go home, perhaps a reluctance to make real what was happening. This was evidenced as Hazel went home. Nothing had changed, nothing had moved, nothing had altered. The past 10 weeks of loss, suffering and death was not seen at home; everything was still the same as it was before Philip had gone to the hospice. A sense of reverie enveloped the home. The stubborn refusal of the space to match the experience led to a sense of disconnection, an almost dreamlike quality; nothing had changed at home, but everything had changed in their world.

As a family of three, they visited the funeral directors. They had spoken before this visit with Olivia about the funeral process; Olivia rejected a cremation and wanted her brother to be buried, a request that her parents honoured. It was perhaps unsurprising that Olivia opted for this, the option that would keep her brother safe, unconsumed by fire. Jeremy recalled his wonderment at Olivia taking an interest in the whole process. Olivia asked the funeral director if she could have a look inside one of the coffins. Jeremy hesitated only to be met with the funeral director profusely saying *"yes certainly"*. The world of 'death' shows its unfamiliarity with Jeremy's reaction. Having a look inside a coffin is alien to the adult world, something that is uncomfortable and almost feared due to the taboo it represents. Yet for Olivia, her childlike years did not see the fear of death that the world has attached to it:

> Jeremy: "*Olivia looked inside and she ran her hand around the lining and she said 'yeah that's lovely… that'll be just right for Philip."*

This almost innate dread of death and trepidation of corpses seems to develop as people age. Olivia showed no fear, no sign of this being not the right question to ask or act to carry out. Jeremy was trying to protect her, trying to stop her from mixing with death, yet, for her, the comfort and silk lining of the coffin meant that it would be comfortable for her older brother to rest in. The day before the funeral arrived, the decision was made that they did not wish to be present when the funeral directors came to move Philip from his bed into the coffin. The finality of this appeared to be too much; it is as if they wished to still picture Philip lying in a bed, where he was present and accessible, much like the bedroom maintained by Caroline for Danielle. This was perhaps magnified by the opportunity of the preceding six days, where they could go and be with him. Still the weight of responsibility for Philip lingered and before the funeral directors left Martin House, they all went and placed their hands on top of the coffin, letting Philip know that they were with him.

> Jeremy: *"…if you spend… …. Erm… nine years if you like… ….erm…. gradually beating this steady retreat doing your Best to ensure that… he's he's okay and hoping against hope that he will survive…. But if your entire focus is on that then when it comes to the child's death then that is quite difficult to accommodate… erm… so…"*

The years of sacrifice, of caring for their son, the conflict they experienced in themselves in not wanting their son to suffer, but also not wanting to lose him had now been taken out of their hands. The pauses in the above quote emphasised this. The difficulty of discussing this aloud, of accepting again that this had happened, caused Jeremy to tear up as he relived these painful moments. This was emphasised further in the burial of his son, and the loss of physical closeness in flesh of my flesh that was now able.

> Jeremy: *"the… there was the… the things said round the grave and of course then the coffin went in… erm… that is… that is a bit of an ending… …."*

> Jeremy: *"I felt uncomfortable when I came home because as darkness fell ermm.. we'd done our best to look after him and I felt in some ways I was letting him down"*

Distance was profound here. A permanent distance from his son, from his identity as a carer, no longer being able to protect him because he was no longer close by. He was no longer in this world. Darkness came with the evening, but also in his lived experience of confusion. This is evidenced in a poem that Jeremy wrote:

Postscript

All this is but now a receding past:
A history,
enveloped by the swirling mists of passing time;
a fading memory of distant fears and aching loss.

Although increasingly remote,
These past events still lurk in the shadows of the unconscious mind.
Rekindled, in an instant, by any spark of similar circumstance,
they rush to the forefront of everyday thought:
an explosion of raw pain and hurt
carried within a surge of overwhelming sadness.

But, Life proceeds
and Fate continues to wield its sword,
felling, in time, all those we love and hold dear.
For those yet untouched and still in youth,
we must prepare for when we see its glint.
For out of knowledge comes hope
and the power to counter what should not be.

And as for ignorance?
It shuffles hand in hand with despair.

The responsibility of having and caring for a child is strongly felt when being a parent. This sudden ending to the depth of care that Jeremy had given him was difficult for him to cope with in the face of death. The thought of Philip being alone, being cold, being in darkness prevented him from visiting Philip's graveside.

> Jeremy: *"I mean something that I found very difficult was that having spent so long protecting him and looking after him and trying to keep… unpleasant things at a distance… when he was… erm… after the funeral… and he was in the graveyard… coming past it… it was a rainy night… and I felt very uncomfortable about being in the warm house… it all sounds very strange… … but I I I realised… I can understand that now because I felt.. that like… I should still be looking after him… he shouldn't be by himself even though he was dead… he shouldn't be by himself in the graveyard… and it was a real struggle to know… that… I should just… he's buried and I should I should just leave him there."*

Spatiality and embodiment merge together here. Physically visiting the grave made the situation too real, too literal for Jeremy. His son was underneath the ground and alone and he could not do anything about it. The guilt he felt a palpable challenge to the Cartesian split of body and soul. Yet Jeremy felt a lack of interaction with Philip when he visited his graveside. There was no contact, no communication, a detachment from Philip. This absence of interaction was something that he found *"emotionally very very taxing."* He described how he gained understanding as to why people brought flowers and ornaments to their loved one's graveside, something which he had not previously understood. He commented how to aid a sense of interaction, the bringing of a gift, for the giver, enabled a sense of contact and involvement. *"Cleaning the headstone"* is akin to a caring role; the continuation of care although they had departed. Grief receded, helped by volunteering to maintain the graveside with a few other older men who did the same, a scene he described as comparable to the television programme 'the Last of the Summer Wine'. They would work and then break for coffee with their flasks and homemade sandwiches. Being in the graveyard, being in the world of death, helped Jeremy feel more comfortable. The fresh air, the company, the woodland scenery in the distance, the sounds of children playing in the playground of the local school, brought a level of safety and acceptance to his son's resting place.

> Jeremy: *"and half way through the morning cutting the grass they would go to a bench… sit there open the flasks that their wives had made and have coffee or soup and sandwiches and there would be the banter… and I realised for me it was very therapeutic that the graveyard then became quite a friendly place and I was able to work in other parts of it cutting the grass and then gradually work towards the graveside"*

This sense of movement towards understanding and accepting death's reality was aided by being present in the graveyard itself, experiencing the quietness of it where it is no longer feared. The three gentlemen who volunteered to maintain the graveside enabled an at-homeness, where the graveyard could be a place of laughter and lightness. In a gentle way, Jeremy worked his way to his son's grave, his son's death and the acceptance of its implications. This has now evolved to Jeremy finding it *"very pleasant going back there now… it's quite restful".*

The words in the poem seem to summarise much of the narrative and interpretation presented in both accounts of the death experience. The temporal element of the lifeworld is strongly conveyed as is the sense of loss and vulnerability. This loss still lurks and lingers, never fully forgotten. Fading suffering is still felt; still prone to being ripped opened like a slow-healing wound. The final lines of the poem reiterate Jeremy's belief that seeking understanding is vital in coming to terms with pain of death. We are encouraged to prepare for and orient ourselves towards death; to come to terms with a Heideggerian being-towards-death, lest it catch us unawares and cut us even deeper.

Embodied interpretation

The physical loss of a body not responding, of life no longer flowing through, of the personhood no longer being able to communicate or comprehend conjures a strangeness, a desperate desire for it to be shaken and awakened again. Drained of colour, and cold, only the unresponsive shell remains. The body of the person that is held so dear is threatened with its removal and never to be seen, smelt or touched again. It seems that death is not the final separation, but instead, the taking away of the body from its carers, its loved ones, its owners. Time is needed to touch and hold the flesh that will soon not be seen again. To carefully position and make comfortable, ready for its final rest, closing its eyes, joining its fingers and laying them on the chest. Time spent with the body, with the person, is precious. An unrushed experience of lying next to them is preferable, whispering to them the depths of your love for them, rather than feeling the pressure of needing to leave the hospital ward, for someone else to take up the bed. Fear of the dead is not felt here. The taboo of death no longer exists as you come face to face with it. It is not chilling or frightening. Rather the body is mourned over and loved.

Following the removal of the body from the loved ones' physical ownership, thoughts now move to the shell of the body, the person that is still to be cared for. The 'best' for the deceased is now desired by loved ones left. The silk lining of coffins as fingers run through it, are tested in terms of the comfort they can offer for the final resting place. Precious items and belongings that were loved by the person are placed with them as they rest, to aid peace and a sense of at-homeness for the deceased. Parents see this as comforting their children, yet are they comforting themselves? This dynamic relation between body and world, between the living and dead is revealed in the care shown to the laying down of loved ones; in their desire to still comfort and protect them.

The body now removed, resting underneath the ground or cremated, can no longer be touched. Yet on clings the love of those remaining, though the forced separation that death brings shatters the life once known, families find their own way, their way of peace, to stay as close to their loved one as death and life allows. The appreciation of scenic views from the gravestone where the sound of children playing at a nearby school brings comfort to the deceased through the eyes of the loved ones left. The interactions of cleaning the gravestone or of bringing flowers and ornaments to the graveside offers the chance of connection to the physical being that death, decomposition or earth cannot separate. The residue of the person can still be present in daily life. Although unresponsive, they continue to talk, to share life with and thank for the ways in which they have improved their existence.

And afterwards, a reconnection takes place with the world that continued to spin whilst lives came to a halt. New ways of living and paradigms are founded. After death there is still a sense of mobility, but this is stilled in the dark of night, when torturous thoughts hit. Clarity cannot be found in darkness. Yet, the light of day, majestic scenes of nature, the oceans and grandeur of mountains aid in blowing these intimidating thoughts away. Those thoughts that question identity and love; there is a need for something greater and vaster than the situation, a need to embody, to feel, to offer perspective and balance.

As new people walk into the shared horizons, a question is anticipated regarding family. The sense of choking or a lump building in the throat is felt. The questioning of self, which world to bring them into is fractiously determined. The disruption to the lifeworld that illness brought at the beginning of the illness narrative will continue its separation, now through the death of other people understanding you, and you understanding other people. *"There is no light at the end of the tunnel"* [Caroline]. Nothing can make it better and nothing can ease the pain. Rather, a new life to live has to begin. This new life will always feel empty, it was created through loss. Yet new ways of living with the dead are sought, within the legacy of the child lost.

6.3 Discussion

This study has explored the accounts of the death experience of families whose child died of NPC. Adopting a hermeneutical phenomenological approach enabled an exploration of the subjective meanings of the dying experience. The results illuminated the existential vulnerability experienced when faced with the death of a loved one. Conducting an embodied interpretation provided a further level of depth to the understanding of the participants' experiences. This allowed a more empathic presentation of the families' experiences, offering insight into death and dying.

Space was shown to be an important concept throughout the illness and dying experience. For Philip's family, the hospital was seen as a place of imprisonment where quality of care was poor and a lack of understanding from HCPs in terms of care needed was felt. The hospice was a place where death was imminent, but where there was also a sense of well-being in terms of the space evoking freedom; a possibility to step outside and for nature to offer a sense of healing. For Danielle, home was valued more than being in hospital. Although home was a busy place in the final days filled with HCPs' presence, on the last day, a calmness was felt, which created a peaceful dying experience free from the intrusion of others, however liked they were. The *space* in both accounts was crucial to the creation of a context where the taboo of death could be transcended with confidence. In spite of its significance in theory and practice, the notion of 'place' is under-explored within discussions of end-of-life experiences (Lowton, 2009). It is important to factor in such experiences when one considers the fact that most children with a life-limiting illness die in hospital (Cochrane, Liyanage, & Nantambi, 2007; Craft & Killen, 2007).

A Cochrane Review found that for people who wish to die at home, the provision of domiciliary palliative care significantly increases the likelihood of this outcome, demonstrating the importance of specialist palliative community services (Gomes, Calanzani, Curiale, McCrone, & Higginson, 2013). The UK Department of Health End of Life Care Strategy puts emphasis on enabling patients to make choices about place of death and increasing the likelihood of people dying at home if that is their wish (Saunders, 2008). Although this may be what people wish for, only around 20% of people die at home in the UK (Gomes & Higginson, 2008). Vickers et al. (2007) found that parents caring for children who were dying of cancer in the UK stated a preference for a home death and that a large percentage of these patients were able to die at home when key programme components were in place, such as a community nurse specialist team and physician support. (Siden et al., 2008) found that location of death varied by disease, and paediatric cancer patients were the patients most likely to die at home (40.2%) compared to those with more neuromuscular diseases or multi-organ conditions who were more likely to die in hospital. This may reflect the timeline of disease progression and the opportunity to plan for the dying experience either by the use of an advanced care plan or choices over time. It may also reflect the greater availability of more specialist services in more societally prevalent conditions, such as the support given to those with cancer through organisations like Macmillan. Both families included in this study had time post-diagnosis to reflect on and consider what they would want when their children died. However, there were different experiences with regards to the additional help that they received from specialist palliative community services. This demonstrates the inequality of care that people are receiving at this time and the need for community services to be consistent with regards the level and quality of care that they can offer.

The continuity of relationship with the deceased was an important finding for both families in this study. Similarly, Proulx, Martinez, Carnevale and Legault (2016) interviewed bereaved fathers and found that they visited their child's grave often as it was seen as a last tangible link, which brought them a sense of comfort. Proulx et al. (2016) did not give details as to whether this was a process and whether it took time for them to find a sense of comfort at the graveside like it did for Jeremy. However, they found that the interaction of bringing flowers and keeping the graveside tidy felt like a continuation of care for their child. Comparatively, Proulx et al. (2016) found that keepsakes from their children carried a deep symbolic meaning for their fathers and helped them to maintain a relationship with their child. This was the same for Caroline who carried around Danielle's keepsakes with her when she left the house, or had them with her in her bedroom when she was asleep at night.

The breakdown of other relationships, such as friendships, seemed to occur throughout the end-of-life period. The overriding experience of isolation was conveyed by both families, where a distancing from people around them occurred. Van Humbeeck, Dillen, Piers, Grypdonck and Van Den Noortgate (2016) interviewed parents who had experienced the death of their child. Loneliness was described by their participants, expressing feelings of being alone both in their emotional experience and their physical solitude. The current study also illuminated how the breakdown of trust and communication between families and HCPs occurred when significant developments were communicated poorly or when support was not given. This seemed to then catalyse a deeper mistrust of the utility and reliability of supportive relationships with wider friends and families. Both families expressed their negative experiences with HCPs in their lack of empathy and this was a part of the narrative of losing their child, showing the significance these events held within their stories. This highlights the need for HCPs to act with compassion when people are in the vulnerable position of a loved one dying, following the principles of the head-heart-hand in delivering humanised care (Galvin, 2010; Galvin et al., 2018).

Overall, this study explored the existential suffering that losing a child to NPC causes parents. Although both parents achieved their desires in terms of the end-of-life care they wished their children to have, it is still questionable as to how much this would be described as a 'good death.' What constitutes a good death has been researched with varying findings and serves to highlight the difficulty in defining this term (Kastbom, Milberg, & Karlsson, 2017). Overall, the considerations of a good death include pain management, time to prepare for death, feelings of having had the time to live a meaningful life and a quick death compared to a slow death (Granda-Cameron & Houldin, 2012; Steinhauser et al., 2000). Having time to prepare for death was found to be meaningful in this current study. Both families appeared to calmly accept the withdrawing and withholding of further treatment for their children when they had reached a point of care ceasing to be appropriate. This was echoed in Kastbom et al.'s (2017) study who interviewed adults who were living with cancer and found that many of their participants prioritised the importance of preparing for their death and having the chance to say goodbye to their loved ones. This helped them to gradually accept their own death but the relationship between time, suffering and acceptance is not altogether clear. A longer life with more time for acceptance may also lead to greater suffering and slower death, taking the experience further away from a traditional conception of the good death.

Increasingly, attempts to characterise any death as good are shown to be unconvincing. The term 'a good death' seems to be the reserve of HCPs and their considerations. It feels very detached from the actual occurrence of what death is. This study found that both families experienced many aspects of the traditional definition of a good death, such as good support from the hospice and community palliative specialists, dying in the preferred location, surrounded by their family, yet the suffering expressed was very present. Should the dying experience be wrapped within the terminology of a good death? Although the dying experience may well have been made worse by poor care or persistent attempts to prolong life through painful interventions, the final outcome would still remain. Whatever its antecedents, death and its sting still await. Steele et al. (2013) concluded that we know very little about how to improve paediatric end-of-life care within clinical practice and sought to suggest measures that would rectify this. They interviewed parents and siblings of deceased children to solicit their advice on how to improve provision of palliative care. Steele at al. (2013) found that bereaved parents appreciate compassionate care from HCPs throughout their child's illness and for them to not be seen as another patient but as an individual. It is worth noting at this juncture that what is being suggested here is far from revolutionary. To start with the premise that little is known about improving palliative care and to reach the conclusions listed above does not leave us with a richer knowledge of the good death. This is not to suggest that the conclusions of Steele et al. (2013) are incorrect, but that the notion of the good death may be flawed.

It appears from the results of this current study that end-of-life care can cushion the dying experience, but nothing can make that process wholly good or relieve the suffering. Instead, it is an individual experience that will be understood differently. Greater appreciation of the suffering and vulnerabilities of losing a child should be pursued rather than attempting to package the experience within a generic notion of a ‘good death’. Understanding the effects of dying and death from NPC on the lifeworld offers the chance to understand their particular needs and subsequently the manner in which these can be met in specific, targeted ways. It is imperative for healthcare research to focus on deepening understanding of the experiences of families who are bereaved or have a family member dying with NPC. Supporting families in this could help in preparation for death and from there enable end-of-life care decisions that would be most appropriate for their child.

Our modern desires to conceive death as either the problem to be solved, or a distant nuisance to be ignored have led us towards unhelpful framings that only serve to hurt and isolate those in the midst of the experience. The bringing of individuals and institutions into a more honest relationship with death could serve to remind all parties of the intimate and unique nature of death and bereavement. Recognition of the humanity of such experience could be the key to unlocking an honest death through which an individual might have the freedom to meet the event on their terms and be surrounded by communities and institutions that listen, appreciate and understand. The issue of practicality looms large here, as does the question as to what the implications of such conclusions would be. Many attempts to measure or frame the dying experience lead to the unnatural imposition of frameworks onto the individual. It could be argued that the attempt to define or describe is in itself flawed and misguided but to go that far would be to deny any commonality of experience or indeed that we have any values at all in the face of death. What this study suggests is that we may be defining the wrong thing. In a society that is increasingly removed from and shy of the realities of death and dying, can we say that we know enough of these phenomena to adequately conceive of the good or the bad? Starting with a stronger and more coherent conception of death and being guided from there by the unique narratives and experiences of individuals and communities may enable us to value honesty over targets and ignorance.

6.3.1 Reflections

This study provides an in-depth analysis of the death experience of a child with NPC. Exploring the process including end-of-life care, the dying experience and life after death provides the whole narrative, presenting a fuller picture of the experience of losing a child to NPC. Utilising an embodied interpretation allowed for a novel perspective of understanding this whole narrative, in a way that evokes a level of empathic understanding to the experience of both families.

If time allowed, I would have liked to go back to the participants with my embodied interpretation of their narratives to discuss it with them and gather their thoughts surrounding it. This method is a novel approach to research and there are ontological arguments as to how much we can inhabit a body? However, Todres and Galvin (2006) argue that providing an embodied interpretation may help readers understand the nature of a phenomenon in an empathic way by enhancing emotional intelligence rather than giving knowledge. Compared to the other studies in this doctoral research, conducting an embodied interpretation brought a deeper understanding of the data that would not be there if this process had not been conducted.

6.3.2 Conclusion

This study has illustrated the existential impact of the dying experience on the lifeworld with regards to suffering and has explored death conceptually in the modern era. The results have illuminated the need for recognition of the experience in our understanding of death through its different elements, particularly spatiality and its intimate links to temporality and identity. The implications for clinical practice include an honest recognition of suffering and vulnerability during the death experience, divorced from unhelpful notions of a ‘good death’. This suffering has been demonstrated to co-exist with concurrent experiences of well-being.

Chapter 7 – A novel approach to scale development using phenomenology: A reflexive approach

7.0 Introduction

The initial aims of this study in accordance with the funder's brief were to develop disease-specific QoL scales for people living with NPC. As my research progressed, I became convinced that the assumptions inherent in more traditional models of scale development would not fully capture the nuances of living with NPC. Therefore the decision was taken to explore novel means of scale development as an alternative, seeking to take seriously the different dimensions of the lifeworld and a phenomenologically-informed conception of QoL. Led by a phenomenological worldview, the previous empirical studies in this thesis explored in-depth the experiences of living with NPC. I will now move on to reflectively consider how I conceptualised and operationalised the concept of 'quality of life' from the subjective experiences of people living with NPC gathered from these series of studies in order to develop QoL scales. In this chapter, I will firstly revisit the concept of QoL and its utilisation in terms of patient reported outcome measures and provide a rationale as to this novel approach to scale-development using phenomenology. I will then outline the developmental stages used to develop two age-appropriate QoL scales for people living with NPC based on the participants' experiential narratives of living with illness. This chapter will end with a reflective discussion on the use of QoL scales within a clinical setting for people living with NPC and future recommendations of this.

7.1 The conceptualisation and operationalisation of QoL re-visited

The concept of QoL is well-established in both research and clinical practice. This is the case in spite of the lack of consensus on the definition (Kassianos, 2015), (see chapter 1.3 for a more detailed exploration of the concept of QoL and instruments to measure this). Quite often in research, papers looking at the concept of QoL do not provide a clear definition of what it means within the context that it is being used and in doing so, assumes the reader's understanding (Cohen & Biesecker, 2010). This lack of clarity makes the conceptualisation of QoL and the operationalisation of it within measurable instruments inconsistent and questions arise as to its precision (Anderson & Burckhardt, 1999). Patient reported outcome measures are often used in clinical trials to assess a person's QoL, providing evidence of the effects of interventions from the patient's perspective. Their goal is to effectively bridge the gap between how healthcare professionals perceive their patients versus how their patients experience illness and understand their QoL (Fitzpatrick et al., 1992; Jacobs, 2009). This strive to promote a person's well-being by invoking QoL as an outcome measure is problematic when the concept of what constitutes QoL has not been clarified or when it is not explicit as to how researchers have reached their working definitions. Arguably one of the most respected definitions of QoL is the WHO's definition, which defines it as being multi-dimensional, taking into account one's physical health, psychological state, and social relationships making these the key domains included in outcome measures when measuring QoL (Kassianos, 2015).

Ferrans and Powers (1992) presented a different focus on what QoL means in that they argued it is dependent upon satisfaction or dissatisfaction with areas of life that are subjectively deemed important. This definition of QoL seems to offer a limited view of the complexities of human existence by focusing on the notion of satisfaction. Indeed, considering this from a Heideggerian outlook, anxiety around us moves us towards an authentic existence as we live in a way that is oriented towards-death (Sarvimäki, 2006). Therefore, feeling dissatisfied could indeed orientate a person towards a better understanding of what living authentically entails, thus provide them with a quality of *being* that is more grounded. The series of studies in this thesis demonstrate the nuanced, deep-rooted intricacies that living with illness has on the lifeworld, enwrapped within both well-being and suffering. The reduction of the concept of being to notions of satisfaction or dissatisfaction seems almost belittling to the complex narratives that have been told (Benner, 1985; Draper, 1992). This therefore provides an argument for a richer and more nuanced conception of QoL, especially when living with a LLC.

With a view to developing outcome measures that assess QoL for people living with NPC, looking through the lens of traditional QoL definitions at the complex experiences of living with a LLC shown in the previous chapters, questions arise as to whether previous definitions such as the ones mentioned above, enable an exploration of QoL that take on board these nuances. Such a critique is aligned with the Heideggerian approach taken in this thesis. Heidegger (1978) criticised the concept of a fragmentation between humans being made up of three parts: body, mind and spirit. For Heidegger, being human is *Dasein*; this is the basis of our understanding of ourselves (Large, 2008; Shariatinia, 2016). This is not a distinct physical, observable entity, but the construction of a life's narrative (Sarvimäki, 2009). With this in mind, it was important for the scope of the developmental process of QoL scales to be formed on the basis of one's narrative of living with NPC, focusing on the subjective meanings of living with illness from those who are impacted the most. Garnering this from the lived-experiences allows for a more detailed understanding of what Quality of *being* encompasses.

7.2 Phenomenology as a novel approach to scale development

The rationale for the use of phenomenology in this study over more traditional scale development methods is to give value to the lived experiences and sense-making that occurs when people are living with illness. The relationship between phenomenology and individual's self-perception was explored by Merleau-Ponty (1945/1962) who spoke of the objective body (*le corps objectif*) and the lived body (*corps propre*). When illness is not present the two bodies live in harmony. This changes when illness comes and the objective body becomes centre stage as it ceases to function in the ways to which one has become accustomed. As Carel (2011) said the body's "dysfunction is so important, so intimately linked to our wellbeing because it is us. Illness is a painful and violent way of revealing the intimately bodily nature of our being" (p.40).

For children and adults living with NPC, as shown in chapters 4 and 5, we saw the change in the things they once could do to the things they could no longer do. Illness robs the individual of the ability to engage with everyday actions (Carel, 2011). Thus revealing the divide between the objective body and the lived body in not only the lost physical ability to walk or hold toys, but the meanings and experiences that are behind these things. This is especially pertinent to determine when we are considering firstly an illness that is incurable and secondly, one that is rare. In order to develop a disease-specific scale, it is necessary to explore in-depth the existential issues that come with an illness with no cure and one that is unrecognised and underappreciated by healthcare professionals and wider society. Thus, this encourages a movement away from the reliance on the traditional compartmentalised QoL constructs, with an attempt to conceptualise QoL and transcend the physical, mental, social and spiritual in order to deepen understanding of the meaning of living with NPC. Accomplishing this necessitates an engagement with broader philosophical constructs and frameworks that can be used to understand such phenomena.

7.2.1 Setting up the project

The series of studies in this thesis have been underpinned by phenomenology as a framework for understanding people's experiences of living with NPC. Working from an interpretivist phenomenological paradigm meant that the phenomenon of experiencing NPC as well as illuminating more deeper-rooted, internal meanings of the illness experience has provided a contextual and informed understanding of the experience. The strategies chosen to guide the epistemological axis of the series of studies have meant that the emphasis has been on the *corps propre* of NPC as opposed to the *le corps objectif* experience, a distinction which is an important feature of phenomenology (Carel, 2011). Galvin and Todres' (2013, 2011) lifeworld theory has also been used to underpin these studies in order to broaden the scope of understanding the illness experiences on a more fundamental and holistic level as to how human beings understand the world about them. Seeing this from a QoL perspective, it could be argued that many of the principles within pre-existing QoL domains complement the lifeworld-led care constructs. Yet I would argue that the lifeworld-led theory allows for a more open, less constrained view, which incorporates the shared and unique dimensions of individuals' experiences that are central to their QoL; something which is particularly important when looking at a rare disease about which we know very little (Todres, Galvin, & Dahlberg, 2007).

In this chapter, I will shift focus to consider the depth of what constitutes life's *quality* from an ontological perspective using data gathered in chapters 4 and 5 for people living with NPC. As Bunge (1975) argued: "A better understanding of the quality of life calls for more intense theoretical and methodological work rather than an increase in the amount of social and environmental statistics… data without ideas are sterile when not misleading" (p. 65). With this in mind, arguing against a hedonic conception of QoL with relation to Ferrans and Powers' (1992) definition, this study instead aligns itself with the Heideggerian notion of *Dasein* and the needs, priorities and meanings of people living with NPC, abiding within the interpretivist paradigm that guides this thesis. This means recognising a notion of *quality* of *being* that is intricately linked to common experience, to the objects that are interacted with and the people that are known. This is understood in conjunction with an orientation towards the future, towards death, towards the completion of life's narrative. This study therefore interpreted 'quality' as being that which helps or prevents people from living an authentic life using the description related to Heideggerian understanding above. Any disruptions to this were explored and seen as a key avenue of exploration in how living with NPC may impact upon a person's quality of being. A sense of being-at-home was seen as experiencing quality of life, going further than satisfaction or dissatisfaction. Arguably a stronger emphasis on the developmental process may provide a more robust basis for QoL scales based on lived-experience in conjunction with theoretical models that aim to provide the fullest explanation possible as to what it is to live with illness.

Therefore this current study, underpinned by phenomenology and the lifeworld theory, sought a 're-analysis' with a new purpose and different objective, seeking to develop items for two age-appropriate, disease-specific QoL scales for people living with NPC using the findings reported in chapters 4 and 5. Developing a tool informed by a lifeworld philosophical orientation will provide HCPs with a better understanding of what it means to live with illness. This could also give clinicians a better grasp of their patients' experiences as opposed to purely focusing on the disease trajectory in such a way that fails to transcend reductionist specialisms.

7.3 Method

7.3.1 Design for scale development

An explanation-driven logic of inquiry was adopted which aimed to conceptualise ‘quality of life’ from the subjective experiences of people living with NPC, with an exploratory analysis rooted in phenomenology. The lifeworld theory was used to further deepen the analysis, producing the best possible explanations as to the effects of NPC on all aspects of QoL, drawing upon the disruptions to the elements of the lifeworld and how these may impact upon an understanding of QoL. The initial phenomenon under exploration in chapters 4 and 5 was living with NPC as experienced by people with a diagnosis and their parents or carers. How QoL is impacted upon when living with NPC is the specific focus for this study. As previously mentioned, this study interpreted ‘quality’ as being that which helps or prevents people from living an authentic life. Reflexive practice was also incorporated as this further disclosed the lived world of NPC and its impact on QoL. The proposal for developing QoL scales was explained in the same participant information sheet as used in chapters 4 and 5 (see Appendix 3) and therefore was covered by the favourable opinion given by the Research Ethics Committees (ethics number: 15/WM/0093).

7.3.2 Participants

As reported in chapters 4 and 5, fourteen biographical interviews were conducted with people diagnosed with NPC and/or parents or carers of people living with NPC. Eight parents/carers of children were interviewed and 6 parents/carers of adults (see sections 4.1.3 and 5.1.3 For participant information).

7.3.3 Data collection

As reported in chapters 4 and 5, an advertisement for the study was published on the NPUK Charity’s social media pages and interested participants contacted the researcher. The interview took an in-depth biographical approach to data collection in order to provide a more natural narrative of the lifeworld that is led by the participant as opposed to the interviewer. Overall, participants were asked to talk about their experiences of living with NPC. The schedule was not prescriptive or inclusive but acted as a guide in the interview in order to ensure the aims of the study were met. Interviews were recorded using a Dictaphone and transcribed verbatim.

7.3.4 Qualitative analysis of interviews

In order to ensure the items for the QoL scales were age appropriate, data were analysed in two age groups, children with NPC 0-17 years and adults with NPC 18 years and over. The interview data as reported in chapters 4 and 5 were analysed using IPA. Focusing on the biographic accounts of people living with NPC and their parents'/carers' meaning-making of the experiences enabled a further, more in-depth exploration and understanding of the phenomenon of living with NPC.

For the scale development process, transcripts were re-read in order to familiarise myself with the data becoming attuned to the new focus on semantic content and striking language used by the participant, which may be related to the impact of NPC on their QoL. The themes generated through IPA were the starting point for this analysis. The phenomenon of focus in the original analysis was the experience of living with NPC. In this analysis, the goal shifted to focus in on the concept of 'quality of being' with a particular emphasis on what helps or hinders people in living an authentic life. The data were interrogated against the elements of the lifeworld to identify how disruptions to the lifeworld may impact on life quality. Themes and sub-themes were explored in more detail by going through each element of the Lifeworld: Temporality, Intersubjectivity, Embodiment, Spatiality, Identity and Mood, using the lens of living with NPC and the disturbance or impact this has had on the lives of those diagnosed.

This involved a cyclical dialogue between the primary inductive themes generated alongside the elements of the lifeworld, thus embedding the inductive analysis within existential-phenomenological theory. Also involved in this cyclical dialogue were notes from my reflexive journal made after the interviews with participants when thinking about how NPC had impacted upon a person's QoL. This took an embodied interpretation approach which complements phenomenology in valuing the embodied understanding of being human. Over the course of this thesis, I have gleaned information about the experiences of living with NPC in ways that go beyond verbal accounts. Interacting with the NPC community for the past four years has meant that I have seen deterioration, frustration, contentment and distress regarding the illness experience. This differs from the medicalised perception of the objective body, going further than the ataxia presented or the wheelchair. I have documented these moments along with many others during interviews and other encounters which are based on my perceptual experiences. Amidst the dialogue with inductive data and in conjunction with the lifeworld theory, incorporating these reflective moments into this scale development was important to the research. Discussions were held within the supervisory team throughout all phases of analysis so we could be confident that the interpretation of the data produced was the best possible explanation of QoL when living with NPC.

7.3.5 Item generation

Due to the neurological effects of NPC on patients, it was decided after conducting interviews that it would be more appropriate to develop all the scales as proxy scales, rather than for direct use with the patient as very few people with NPC would be able to answer a QoL questionnaire (Carlozzi et al., 2016). From the cyclical process between the IPA analysis, the lifeworld theory and the reflexive contribution, a bank of all possible items, which were considered to have an effect on an individual's QoL were noted. This involved the construction of a matrix of all reported themes and sub-themes with descriptive summaries/phrases relevant to QoL set against the elements of the lifeworld. Themes and lifeworld elements were cross-compared to produce a list of all potential items that could be used to describe the QoL of someone with NPC. Variations of similarly-focused items were worded differently and were kept on the list to establish which item was worded the most appropriately. Examples of items in relation to the lifeworld elements are reported in Table 7.1 for children and Table 7.2 for adults. These possible items were then discussed within a team of psychologists, notably my principal supervisor who is an expert in scale development and validation, as well as several meetings with stakeholders who work with NPC patients and families. From these meetings, similarly-worded items were omitted as a result of this discussion.

A mixture of closed questions and open-ended questions were developed for the child and adult scales. The open-ended questions were intended to garner a richer perspective on any additional needs of the person with NPC; these questions were optional. All closed items were written as statements with a 5-point Likert response scale from 1 (never) to 5 (always). As the scales were developed as proxy versions, parents/carers were reminded in the instructions to answer the questions from the perspective of how they felt the one they were caring for would answer; this questionnaire was not assessing their experiences, but the experiences of the ones for whom they were caring. The final child prototype scale (NPCQLQ-C) consisted of 28 items; the adult prototype scale (NPCQLQ-A) consisted of 38 items.

7.4 Results

7.4.1 The Niemann-Pick type C Quality of Life Questionaire – Child (NPCQLQ-C)

The NPCQLQ-C items focused on the embodied experience of living with NPC, both in terms of the objective, physical manifestation of NPC, and the subjective experience of contentment within their lived body. Items concerning bodily alienation, in terms of the disconnect from the body once known were shown to be important to a child's QoL. This was demonstrated in the frustration no longer being able to take part in activities that they could previously, such as holding toys or using apparatus in the park. Items concerning space and disruptions to the home environment were deemed to be important in that these changes were made to compensate for the lack of mobility or the anticipated bodily deterioration that would come in the near future.

Table 7. Matrix of themes, lifeworld elements and example items for the NPC quality of life scale for children

Theme	Element of the Lifeworld	Reflective notes	Items for a QoL scale for children with NPC
Learning to live with the clinical manifestations of NPC	Temporality Embodiment Mood	Present mobility compared to past. Recognition of the rate of deterioration. Extent to which NPC has manifested on their physical being and the implication this has on how they feel about themselves and their future. The felt sense of bodily self-deterioration. No longer able to do what they could before. Adjustment to physical deterioration and the possibilities that equipment and support care staff bring.	My child feels content in day to day life My child feels sad because of NPC My child looks forward to new experiences in the future My child feels comfortable with their body
Relationships as security and enablement	Intersubjectivity Embodiment Mood	A felt sense of belonging provided by family and friends giving a sense of peace Continuity of care and recognition of healthcare professionals and support staff offering a sense of familiarity.	Family time is important to my child My child feels supported by siblings/friends My child sometimes feels alone Being in company with others is important to my child My child feels similar to other girls and boys their age
The continuous journey of adaptation creating opportunity	Embodiment Identity Intersubjectivity Spatiality Mood	Adjustment and utilising equipment. Making changes/amendments to increase or sustain freedom. Feelings of loss identity and journeying towards an acceptance of their new sense of self.	My child feels different to other children My child enjoys new social experiences and environments My child copes well when things change at home The home space helps my child feel at peace

Table 8. Example of items included in the NPCQLQ-C

	Item		Item
1.	Because of NPC, my child's mobility has been affected	9.	My child enjoys spending time with their friends
2.	Because of NPC, my child struggles to hold things in his or her hand	10.	Family routines make my child feel secure
3.	My child enjoys playing with their toys or equivalent	11.	I think my child feels like they cannot do things other children their age can do because of NPC
4.	My child feels content about their physical health	12.	I think my child feels like their relationships with their friends have changed because of NPC
5.	My child feels comfortable with their body	13.	Changes to our home environment have made day to day living easier for my child
6.	My child is excited by the prospect of taking part in special occasions	14.	My child appears to be sad
7.	My child experiences times of frustration regarding their symptoms of NPC	15.	My child copes well when things change at home
8.	My child generally enjoys daily life		

7.4.2 The Niemann-Pick type C Quality of Life Questionnaire-Adult items (NPCQLQ-A)

The NPCQLQ-A items focused more on the intersubjective sense of self and how this related to the temporal element of the life they once knew before their diagnosis with NPC, compared to the life they are now living. Many of the adults diagnosed with NPC had to leave employment and their social relationships had changed. This impacted the mood and sense of identity linked to the fact that many participants interviewed had previously had a career before their diagnosis. The past was something that they longed for and the future was something that felt daunting and hopeless. This change in identity led to spells of anxiety and depression due to the life they felt they had lost. Although many of the adults had better physical health than children with NPC, there was a sense of burden in the illness they were carrying, thus the impact on QoL could be greater, highlighting the complexity of the effects of this life-limiting condition on a person’s QoL and how this is not necessarily directly associated with the physical/objective body and the severity of disruption to this. Relationships had suffered for all of the participants, communicating feelings of being let down by old friends and not having the confidence or even the inclination to form new friendships. Without explicitly exploring each element of the lifeworld, these nuances in people’s experiences may not have been identified.

Table 9. Matrix of themes, lifeworld elements and example items for the NPC quality of life scale for adults

Theme	Element of the Lifeworld	Reflective notes	Items for a QoL scale for adults with NPC
Variegated experiences of loss	Temporality Spatiality Intersubjectivity Identity	Past was full of potential with regards to life goals. The present now comprises of loss and the future is no longer anticipated with a sense of excitement. It is instead seen as hopeless. Loss of self-esteem and independence. Reliance on others. Loss of careers and progression, which affected how they saw themselves and their sense of identity	The person with NPC feels confident in who they are as a person Because of NPC they are not able to do the things they would like to do On the whole, they feel content They feel more content at home than in other settings
The experience of diminishing control	Temporality Spatiality Identity Embodiment	Pre NPC diagnosis, felt in control. Now learning to cope with new body that they no longer feel in control of. Feelings of disconnect from society. Feelings of embarrassment at not being able to control body in society.	The person with NPC feels in control of their present They feel in control of what their future looks like They feel they are the person they want to be They feel they want to hide away from the world
Longing for social meaning	Embodiment Intersubjectivity Temporality Identity	Comparison of what life was like pre-diagnosis. Lack of hope for the future in finding friends/close relationships. Struggle to relate to others – no shared experiences. Feelings of isolation and loneliness.	The person with NPC feels disconnected from society They feel that they do no relate to other people They feel that being with friends takes their mind off things that they find difficult They feel that they have a lot of things in common with their friends
Hiddennes, perception and identity	Temporality Intersubjectivity Embodiment Identity Mood Spatiality	Rare and varied disease; struggle to find others to share experiences with. Shell of body – looks healthy but body lets them down. Feelings of embarrassment around the cognitive manifestations of NPC. Limbo land: unsure what their identity is. Separates disability from self, their body does not belong to them, imposter.	The person with NPC feels that they are the person they want to be They feel that things in life are meaningful to them They feel motivated every day to achieve what they want to achieve

Table 10. Example of items included in the NPCQLQ-A

	Item		Item
1.	The person with NPC feels supported in daily life	6.	They feel content with their body and how it works
2.	They feel hopeful when they think about the future	7.	They feel motivated every day to achieve what they want to achieve
3.	They feel that NPC has affected their social interactions	8.	They feel that their body does not allow them to do things that they want to
4.	They feel that they do not relate to other people	9.	They find it easy to make new friends
5.	They feel isolated	10.	The person with NPC feels content when they think about the past

7.5 Reflective Discussion

Adopting a phenomenological approach to explore the experiences of living with illness allowed for a richer understanding of how QoL is impacted upon when living with NPC. Using the elements of the lifeworld enriched our understanding of participants' subjective experiences, which then ensured the items included in the scale represented participants' worlds when living with NPC. Furthermore, drawing on the lifeworld theory meant that both measures, the NPCQLQ-C and NPCQLQ-A are theoretically underpinned by an existential-phenomenological theory, which transcends the individual. This has gone beyond the concepts of QoL used in more traditional measures, adding to the novel approach to operationalising QoL. The intention of such a design is to prioritise the concerns of the individuals involved by first exploring the nature of their experience and moreover, to examine the meanings those people attribute to their experiences. The items above were developed to capture the experiential, real-life impacts of NPC on the daily lives of people diagnosed. This is especially valuable when working with such a rare condition because it then means that these meaningful experiences, rather than preconceived notions from other QoL scales, are used as the starting point for a measurement that can then be used in clinical practice and research. The scales have the potential to be meaningful within a clinical setting in helping healthcare professionals have a richer understanding of the illness experience for people living with NPC. The fact that these scales are designed for a population that is under-researched and often unidentified means that these disease-specific scales are the first of their kind. This novelty is also seen in the methodology used to develop them, which has sought to incorporate underrepresented considerations in understanding such experiences.

As mentioned earlier on in this chapter, QoL scales can be used in clinical trials to provide evidence from the patient's perspective on the effects of an intervention. Both the NPCQLQ-C and NPCQLQ-A have the potential to be used to see if an intervention has affected the person's illness experience in a more meaningful way, providing information for patients, parents/carers and healthcare practitioners on the effectiveness of drug treatments and other health-related interventions for those living with NPC. However, as NPC is a rare disease, clinical trials are few and far between. For example, from personal communication with the NPUK charity, approximately only 20% of the population diagnosed with NPC in the UK were enrolled onto a clinical trial in November (NPUK, 2018). Therefore, it is important to consider how best to integrate the use of these scales in a way that benefits all people living with NPC as opposed to the few who are enrolled onto a trial. These issues are pertinent and meaningful for the successful and practical development of scales to be used in practice. Though, prior to further exploring the potential operationalisation of these scales, I think it is important to consider again the concept of QoL, the connotations it holds and the labels that are attached to these scales.

I think one of the main reasons why I needed to move beyond the scope of QoL and focus on a more holistic experience of living with NPC when embarking upon this programme of work (explained in chapter 3.1.2) was because of the word 'quality'. The word 'quality' suggests excellence, something that is good or something that can be improved upon when measured against something else. The progressive and degenerative nature of NPC and the experiences of some of the participants I interviewed seemed to go against the connotations attached to this word. The notion of 'quality' of life is difficult to ascribe when living with a LLC. Anecdotally, I had a number of discussions with people who had cared or who were caring for a family member with NPC. As a way of gaining face validity to the items developed from this study, I conducted cognitive interviews and showed several people in the community the relevant prototype QoL scale, explaining that these scales were being developed to measure a person's QoL when living with NPC. Some reservations were conveyed in that some believed that they would not have been able to answer a few of the items, as they would not have known how the person they were caring for felt.

Of course, in order to enhance understanding in a real-world setting, more could be done to make sure the instructions given at the beginning of the questionnaire are clear and that they encourage people to think beyond the objective ability of the person and take into account the intricacies of non-verbal and embodied communication. The wording could seek to give emphasis to subtler forms of communication such as changes in facial expressions or mood. However, it is worth considering that some of the reservations held may be due to the connotations attached to completing a 'quality of life' scale, detracting from people's ability to answer in a way that feels 'real' to them.

As discussed at the beginning of this thesis, I was seeking to understand the experiences of people living with NPC through the lens of 'quality', which prevented me from getting to understand the complexities of the NPC experience. This may be similar to people who could potentially be asked to complete one of these QoL scales in that they are seeing their experience through the lens of *quality* of life which may impede upon the ways in which they connect with the items. From the results of the empirical studies presented in this thesis, we know that people living with NPC experience both well-being and suffering (discussed more in chapter 8.2.3). The recognition of this in people's stories gives value to suffering in a way that the word 'quality' does not. I would argue that labelling the above scales as 'illness experience discussion aids' may generate a different perspective and thus a different approach when completing one. This change in language away from QoL recognises illness as the temporal experience it is; an important priority for the NPC community.

I think it is necessary to recognise at this juncture that the shift of emphasis away from the language of ‘quality’ does not mean that I believe life cannot improve for those living with NPC or indeed that interventions should not focus on aiming to improve the experiences of people living with LLCs. Rather, I am arguing that the perception of *quality* is not the same as the perception of illness and in order for intervention to be effective, surely this is what we need to grasp more clearly. The simplistic dichotomies of good or poor, happy or sad, which are often associated with the word ‘quality’, may be united by closer attention to the experience of the person in question. A discussion aid may also realign it with the idiographic illness experience as well as the wants and needs of the patients. It may bring a more fulsome understanding of the person’s story, which is more holistic than quality alone. This is not to suggest that QoL measures are always inappropriate, indeed in some contexts they work well, especially in chronic diseases. Yet from the results of this programme of work, the nuances of living with NPC have been unveiled through understanding the lifeworld and starting from this base seems like a better way to understanding living with NPC holistically than purely focusing on quality. Effective care can be built on this kind of foundation; a robust understanding of the lifeworld and the illness experience (Toombs, 2001).

Todres, Galvin and Dahlberg (2014) argue the value of the “insider” perspective within a caring environment. They describe this ‘dimension of our humanity’ as the: “‘soft underbelly’ that often lies hidden in the shadows. It is both the place that hides our vulnerabilities and therefore often the place that is neglected in our discourses.” (p.9). This perspective is a dimension in Galvin et al.’s (2018) humanising care theoretical framework in prompting HCPs to: see the person behind the ‘illness or condition’; ‘provide care that is not only technical/task focused’; show willing to ‘want to know about patient’s fears about their situations’. By providing care that is attuned to these existential issues, it becomes possible to get to the heart of what really matters to people when living with incurable illness (Galvin et al., 2018).

The tension between the worldview that prioritises the healthcare system and the worldview that prioritises the lived experience of illness, broadly those of the doctor and patient respectively, has been found in previous research (Dahlberg, Todres, & Galvin, 2009; Mishler, 1984; Walseth & Schei, 2011). Within the context of a care conversation, these opposing lifeworlds are brought into conflict and the dominance of the formal, system-led approach can lead to patients feeling unheard. In the studies within this thesis, we saw a series of encounters between HCPs and participants. One of the most poignant cases were the exchanges that Caroline spoke about in chapter 6.2.3. These left her feeling that HCPs had no regard for the pain she was feeling about her daughter's NPC diagnosis, emphasising the divide between the world of the well and the world of the sick that exacerbated this separatedness. Todres et al.'s (2014) concept of the insider perspective provides a potential approach of care which recognises and 'reaches out' to the patient's vulnerabilities. Caroline's insiderness was not recognised making her isolated and disconnected, directly impacting her sense of being-in-the world. Other studies looking at HCPs appreciating and reaching to understand patients' insiderness have shown that a sense of recognition of the struggle from HCPs leads to patients feeling closer in the relationship, more comforted and more in control (Lundqvist, Nilstun, & Dykes, 2002; Nordgren, Asp, & Fagerberg, 2008).

Todres et al. (2014) argue that HCPs need to be aware of the depth of power that they carry in such situations, wherein such power can be highly destructive. This was evident in Caroline's story in chapter 6.2.3, where over a decade later, the comments on interactions with the consultant and other HCPs illustrated the hurt that has been carried as well as the collateral damage to the family's lifeworld across time, leading to a sense of abandonment. HCPs gaining adequate understanding of insiderness would give more recognition to the whole of the illness experience, the core issues as opposed to merely focusing on the facts of disease; something that is incredibly meaningful to people in this study. The notion of 'anticipatory care' as expressed by Boss (1963) defines the requisite approach in order for HCPs to access and engage with the insiderness of a patient (cited in Todres et al., 2014). Whilst HCPs may never have the time nor the ability to truly know the inner lives of those they treat, seeking to anticipate and imagine their concerns and priorities allows for care that is oriented towards lifeworld sensitivity (Todres et al., 2014: p.9). This existential awareness is vital in the provision of care that meaningfully engages with being human (Todres et al., 2014). With this in mind, these 'illness experience discussion aids' could be a tool for guiding HCPs to engage on a more existential level with the challenges of living with NPC and what this means for a patient's major or minor life projects as well as the vulnerabilities shown in the suffering (Todres et al., 2014).

These considerations dovetail with the work of Habermas who emphasised an empathic mindset towards those in need of care. The series of studies leading up to this point has highlighted the significance of how living with NPC may affect the person's sense of self and bodily identity rather than purely focusing on the physiological impact (cited in Walseth & Schei, 2011). The proposed discussion tools would therefore work to introduce into clinical encounters concerns other than those dictated from a medical perspective. It is through such lifeworld-informed encounters that a richer dialogue could emerge, a dialogue rooted in communicative action that gives voice to all parties involved (Walseth & Schei, 2011). As elucidated in the work of Todres et al. (2014), this is not to be understood as a process of reaching a goal or end point of full understanding, rather the concept of seeking to access the 'insiderness' of an individual is an ongoing process. This process must constantly be re-engaged with in order to correctly position the relationship between HCP and patient. By utilising language, argumentation, justification and questioning, HCPs could be better equipped to create mutual trust and respect as the basis of care (Schei, 2006). In relation to the items developed in this chapter, these 'discussion aids' could help HCPs gain knowledge about the nuances of the illness experience, combined with the HCPs' general knowledge of the manifestations of NPC. This combined understanding as a foundation of care is pertinent for people living with NPC and would be especially useful to have in place to make care decisions, especially with regards to end-of-life care.

The recognition of the illness experience is something that needs to be enacted in a way that moves beyond mere tokenism, especially within the rare disease group. The way we use words and phrases within this population in relation to health and illness needs to be carefully reflected on in order to provide humanised care (Galvin et al., 2018). The phrase 'living well' represents common rhetoric when seeking to conceptualise living with illness. However, with regards to LLCs, this can be an inauthentic phrase for people that can even be perceived as patronising, putting pressure on the person living with the condition to 'live well'. A recent suggestion made from a person living with dementia was to change the phrase, 'living well' to 'living as well as you can' in order to accept the varied experiences people have when living with a LLC, experiences where there is often no easy answer or solution (Mitchell, 2018). There must be space for the expression of suffering. In the midst of our British culture, which is renowned for a particular brand of stoicism, words and concepts such as, 'good-death', 'quality of life' and 'beating cancer' are increasingly used in healthcare, perhaps leading to the ignoring of the issues of the pain of suffering. Some may prefer the positive message of QoL, yet anecdotally, my experience of attending the NPC conferences and talking to families leads me to the conclusion that the overall cry is for recognition of the experience because currently NPC is hidden away from society's gaze.

With regards to both the NPCQLQ-C and NPCQLQ-A, where do these reflections fit? The items in the scales reflect the co-existence of suffering and well-being. They contain items that are based on the homelessness created by the impact NPC has had upon the lives of my participants and they also reflect a homecoming in *Gegnet*, an authenticity in their dwelling-mobility. Items reflect the experiences of both the objective (*corps objectif*) and lived (*corps* *propre*) body, the bodily alienation incorporating the disruptions illness has on relationships, our sense of space and time. These items take on board the transformative impact that illness has on the dimensions of a person's life and experience; how life has changed and whether this disruption is something that is pertinent to the person living with NPC.

Clinically, I think the scales have the potential to provide a dialogic aid whereby existential concerns can be identified and discussed. The phenomenological rootedness of the items may enable HCPs to understand the existential and embodied nature of illness. These items are formed from a deep appreciation for the impact of illness on everyday life, not as a secondary effect of the biological disease but as a primary phenomenon. In practice, the NPCQLQ-C and NPCQLQ-A could provide more of a biographical assessment of the illness experience with a focus on how people living with NPC construct their experiences over time and the challenges and triumphs that come with this co-existence of well-being and suffering. The operationalisation of both is an attempt to move on from this apparently causal relationship between a low score on a QoL scale and poor QoL.

This projects joins others such as Galvin et al.'s (2018) humanising care theoretical framework in equipping HCPs to move towards lifeworld-led care that attends to deeper existential issues of the living with illness, moving on from a focus on the objective body. Utilising a lifeworld-led framework for care within which "the vicissitudes of peoples' lives their wellness and suffering can be meaningfully addressed" – this co-existence being something that was a main finding in this study (Dahlberg et al., 2009: p.270). Seeing this from the perspective of an illness experience recognises the complexities and nuances that matter to people, not suggesting an overall score of whether their illness experience is good or bad, but which items are most pertinent at the moment for them and what disruptions could be addressed to improve the situation.

7.5.1 Considerations

These tools build upon research which is integrating phenomenology into clinical practice, moving on from the natural-science foundations of medicine, where disease is constructed in biological terms only (Boorse, 1997). This could begin to bridge the gap between the dialectical struggle of the voice of medicine and the voice of the lifeworld (Mishler, 1984). These scales could provide a way for families living with NPC to communicate to HCPs the broader impact of living with NPC. This would be achieved through a methodology that moves past the effects of disease manifestations and focuses in on how illness affects the lifeworld. As Toombs (1987) argued "Phenomenology does not see the person's experiences as a subjective account of an abstract objective reality, rather it takes this experience to represent the reality of the person's experience" (cited in Carel, 2011: p.44). Thus, these phenomenologically built tools could be used to bring to light the different perspectives on illness and how these could help construct a shared meaning of illness, especially within the context of rare disease.

This may enable people with NPC to feel that they have a platform, informed by evidence, to talk through items that are pertinent to them as opposed to scoring them through the lens of QoL. I would argue that the aim of these scales moving forwards, is to enable patients to feel listened to, aiding as a tool to help them communicate any difficulties they face living with NPC. The tools could help people impacted by NPC to be reflective, which in turn allows for concrete experiences to come to the fore, designed as it is from the on lived experiences of those living with NPC as opposed to the more abstract concepts of QoL. It could also aid HCPs and researchers to understand more about the lifeworld of their patients and the impact of living with NPC on their everyday lives. At the heart of this lifeworld-led approach is a commitment to humanising healthcare, i.e. ensuring that we treat the whole person rather than simply the condition (Galvin et al., 2018). The series of studies leading up to this point has highlighted the significance of how living with NPC may affect the person's sense of self and bodily identity rather than purely focusing on the physiological impact.

I have conducted the initial reliability analyses on these prototype scales, which have demonstrated excellent internal consistency, which suggests there is good reason to test and evaluate their use in clinical practice. However, moving forward, the issues as to whether it is worth going on to validate these scales in the traditional sense, requires further thought. Due to the rarity of the disease, it would not be possible to get a sample big enough to achieve statistical power. However, as a long-term project, using similar conditions could be a way to validate the scales developed. This is something that stakeholders within the NPC population would be interested in doing. However, the overall reconsideration suggested in this discussion of these QoL scales being used more of a discussion aid would warrant future work in working out the best use of these developed items. This is something that needs further consideration and could be explored in future research.

7.5.2 Conclusion

To combat the ambiguity of QoL and in order for HCPs to have a better understanding of what living with NPC involves, the NPCQLQ-C and NPCQLQ-A tools could help patients to communicate how living with NPC has affected their lifeworlds. These tools were developed to capture the experiential, real life impact of NPC on patients' daily lives for the purposes of both clinical practice and research. These scales could provide a focus for: humanising care practices; using phenomenology in practice (Galvin et al., 2018); moving beyond reductionist and narrow focuses and towards a lifeworld-led model of care; giving voice to people impacted by NPC; and promoting more dialogue between patients, parents/carers and healthcare professionals that is grounded in lived experience thereby delivering meaningful lifeworld-led care.

Chapter 8 – Discussion

8.0 Introduction

This discussion chapter will employ the reflexive approach taken throughout this thesis and incorporate some of the techniques used in embodied interpretation to ensure the depth of meaning and significance of participants' stories are represented 'in kind'. It seeks to illuminate the findings of this study in a way that explores the vitality of the results and the impact these have, moving past the traditional ways of writing that can often be "over-sterile" or even "deaden" the significance of the meanings that have been found (Todres & Galvin, 2008: p.569). The contributions that this programme of work has made substantively, theoretically and methodologically will be illuminated. Considerations are given to the implications of the findings for clinical practice, policy and research. The strengths and limitations of the studies will be discussed along with suggestions for future research. A reflexive discussion of my personal account of conducting this research will conclude the chapter.

8.1 Revisiting the aims

The overall aim of this programme of work was to explore the experiences of people who were living with NPC. The impetus for the research was built from a lack of qualitative work conducted with people living with NPC in the UK on understanding the impact of living with this disease. Motivation also stemmed from the request to develop QoL scales for this population. As argued from the outset, it was important to understand the meaning of living with NPC to enable the development of these disease-specific QoL scales; this exploration would be the crux of this thesis. A programme of work was developed to address this aim, which comprised of the following objectives:

1. To explore the lifespan of living and dying with NPC from a first-person experience: children, adults and dying with NPC.

2. To deepen understanding of the illness experience from diagnosis and how it impacts upon the life of the person with NPC and their family members.

3. To consider how data generated from this study could be translated into a tool that measures QoL.

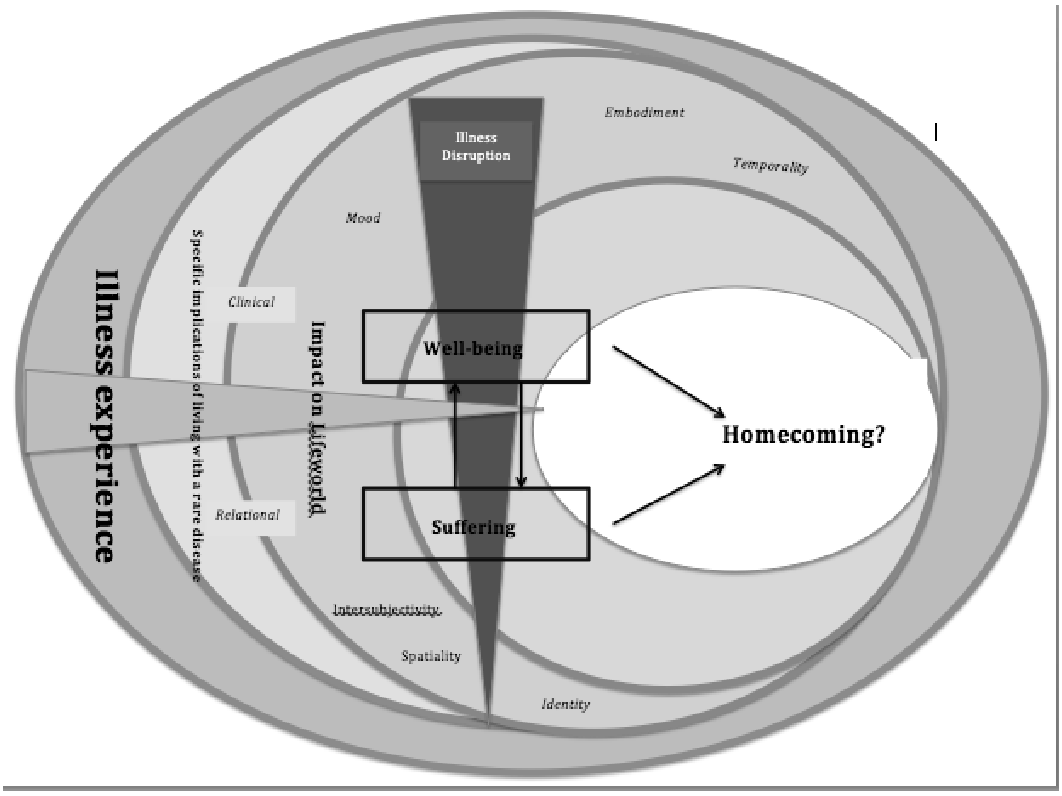
The first two objectives were addressed through interviews with people diagnosed with NPC and with their parents or carers. Adopting a phenomenological framework offered a richer and more nuanced account of life with NPC than may have been achieved using other qualitative analyses, focusing on the phenomenon of living with NPC. Complementing the phenomenological framework, the lifeworld theory was adopted in order to garner the best possible explorations of what it means to live with NPC. The adoption of IPA enabled an idiographic approach to capture the lived experiences of those impacted by NPC (Smith & Osborn, 2003), in line with the aim of this thesis. The hermeneutic arm of IPA favoured the decision to interview parents and carers of those with NPC, to grasp their interpretation of how the person they are caring for makes sense of living with NPC. This provided a fuller picture of not only the person living with NPC, but the impact living with illness has on the family. Gathering experiences and knowledge from the carers allowed for a more layered approach to the narrative of the lived experience of illness. In addition to these methods, an embodied interpretation was conducted to explore the dying experiences of people with NPC. The inclusion of an embodied interpretation allowed me to step further into understanding more about the existential experiences of people with NPC, concentrating on the 'felt sense', permeating beyond words spoken (Todres & Galvin, 2006). This again offered a novel approach, which is underused in health research.

Objective 3 was met through applying the findings from chapter 4 and 5 to develop items for QoL scales for people living with NPC. As described in chapter 7.1, complementing the hermeneutic, Heideggerian outlook that theoretically framed this research, I interpreted 'quality' as being that which helps or hinders people from living an authentic life. The disruptions that illness causes to a person's lifeworld were explored in how living with NPC may impact upon a person's QoL. The conceptual framework of phenomenology and the theoretical basis of the existential-phenomenological theories of suffering and well-being were used in combination to develop items for age-specific QoL scales. This novel approach to scale development generated enriched understanding of participants' subjective experiences, which then ensured the items included in the scales represented participants' worlds when living with NPC. This was pursued in order to broaden the scope of inquiry of more traditional definitions of QoL. Two age-appropriate questionnaires were developed, for children, the NPCQLQ-C and for adults, the NPCQLQ-A. The applications of these scales based on the main findings of this study will be discussed towards the end of this chapter.

8.2 The story of NPC across the lifespan

A synthesis of the main findings will now be discussed; a visual representation of this can be seen in Figure 4. The first layer of my findings is about the specific complexities of living with a rare disease. The discussion will then get to the heart of the matter in exploring the impact that living with illness has had on the lifeworld across the lifespan.

Figure 4. Visual representation of overall results from the programme of work



Following this, the discussion will showcase how the results portray a co-existence of well-being and suffering, which can lead to a sense of at-homeness. In spite of this, homecoming was not experienced as definitive but instead oscillated within the illness experience. Thus, the main body of findings emanates from within the illness experience, which also intersects the other findings, particularly the continuum between suffering and well-being through the disruption to the lifeworld.

The implications that these results have on how the QoL scales (NPCQLQ-C and NPCQLQ-A) could be best integrated in a clinical setting for this population will be provided. Discussions of the implications that the results have to clinical practice will be signposted throughout to demonstrate how closely-related the practical implications are to the substantive claims of the thesis. These implications will be tied together at the end of this chapter and recommendations for clinical practice will be presented.

8.2.1 Living with rare disease

An initial analysis of the findings showed that living under the umbrella of rare disease caused unique complexities and challenges in children and adults who were diagnosed with NPC, along with their family carers. The intrinsic challenges that this imposed upon the participants' lives can be predominantly seen in the clinical challenges interweaved with the impact on relationships, within families and friendships as well as on a more communal level.

Each of the three studies revealed an array of challenging experiences when waiting for the diagnosis. Any delay in diagnosis due to clinical uncertainty can cause concern as people anticipate the outcome and impact that the diagnosis may have on their life. This has been seen in health research looking at cancer where postponed diagnosis leads to experiences of uncertainty (Berterö, Vanhanen, & Appelin, 2008; Thorne, Harris, Hislop, & Vestrup, 1999). Unlike other studies of the diagnosis of rare conditions (Garau, 2016; Limb, Nutt, & Sen, 2010), the duration of uncertainty was considerably prolonged for those diagnosed with NPC. Such experiences were exacerbated by: the failure of HCPs to recognise symptoms; the back-and-forth process of multiple referrals to different specialists; and misdiagnoses being given, which in turn leads to a repetition of the process from the beginning. Diagnoses were quicker if a newborn presented symptoms, yet for many of the families I spoke to, the diagnostic process had taken years and a state of uncertainty showed in that lives began to be disrupted even prior to hearing the words of diagnosis. A lack of trust in HCPs developed due to the length of time it took to diagnose, the way in which the diagnosis was delivered, and the impression that no department or professional stopped the chain of referrals in order to take responsibility for the case; an experience similar to results in Fallowfield and Jenkins (2004). Thus a breakdown between patient-doctor relationships had already occurred prior to diagnosis.

This was happening within a clinical world that did not appear to be knowledgeable or insightful but lacked information about how to help the person in this intermediate state. Knowledge about rare diseases amongst the general public is lacking, but this paternalistically-inspired image of the didactic, expert, all-knowing doctor was crushed by the stark lack of knowledge (Szasz & Hollender, 1956). More specifically, there was a perspective present in the results whereby carers expressed a view of HCPs exhibiting a dispassionate relationship with diagnosis: an irritating puzzle to be solved, rather than a person to be understood (Pinder, 1992). For a mother finding out her son's diagnosis, she described a degree of excitement being shown in HCPs after finally getting it right; a kind of professional pride in the restoration of the natural order (Pinder, 1992). Although the puzzle of diagnosis had been solved, the mother described the limited scope of advice and planning for the following stages. This lack of empathy, or even a lack of interest in her perspective, led her to search the internet in order to gain understanding of NPC. This coincides with other findings where surveys have shown that very few people diagnosed with a rare disease felt that their information needs had been met completely (Litzkendorf et al., 2016) and the internet is often utilised as the main source of information (Garau, 2016).

Litzkendorf et al. (2016) studied people living with different rare diseases along with their relatives. They emphasised the need for fulsome and understandable information at diagnosis in terms of perspective and implications. Contrary to this, I found that on some occasions, families felt that clinicians shared too much information regarding the diagnosis and prognosis of NPC. This overloading of information caused suicidal thoughts for one father who momentarily considered crashing the car after receiving his son's diagnosis, so that they could die together without having to go through the experience of living with this illness. This level of emotion and dread regarding the future showcases the 'narrative wreckage' (Frank, 1995) that the diagnosis of illness can have on the family involved. As well as this, it highlighted their complete lack of thought for what this must be like for the person diagnosed and their parent/carers. The delicacy of excellent communicative skills when giving a diagnosis, seeking to promote honesty as to the implications whilst also providing a supportive environment for people following this, should be recognised as an important component in humanised care (Butow, Brown, Cogar, Tattersall, & Dunn, 2002; Galvin et al., 2018). Galvin et al. (2018) developed a humanised care assessment tool, which uses experiential items informed by phenomenological ideas for HCPs to work with patients to feel human within the world of healthcare. The humanising care assessment tool contains a number of dimensions that attend to 'what it is like' for the person. Galvin et al. (2018) provide eight dimensions of care that contain items to reflect these. From the discussion thus far, the dimension labelled the ‘embodiment-reductionist view of the body’ is pertinent in reminding HCPs to ‘think of the patient as a person and more than just a body’ and to ‘be aware of the physical impact of bad news for the patient’. The dimension labelled ‘insiderness-objectification’ encourages HCPs to ‘see the person behind the illness or condition’ and highlights the importance of being able to ‘show patients that they know ‘what it is like’ from the patient’s perspective’. In addition to this, giving people time and information whilst encouraging people to talk through their feelings are suggested within published guidelines to help clinicians communicate when preparing to give ‘bad news’ (Fallowfield & Jenkins, 2004; Girgis & Sanson-Fisher, 1995).

Research has shown the difficulty that doctors feel when delivering news that people have a disease that is incurable or discussing palliative care options. Studies have found that this sometimes results in clinicians avoiding discussions around the possibility of death unless the patient initiates the conversation. Alternatively, clinicians decide to wait until very close to death before initiating the discussion (Abarshi et al., 2011; Anderson, Kools, & Lyndon, 2013). This is especially the case in ethnic minority groups (Gaveras et al., 2014; Worth et al., 2009). This suggests that clinicians feel ill-equipped to make these decisions and may not know how to manage these conversations for the best (Gawande, 2010). The lack of clarity in the guidelines and the lack of responsibility taken as to who should be having these conversations was discussed in the review of the literature in chapter 2, section 2.3.4. The ‘second conversation’ model (2019), which is currently being piloted by Guy’s and St Thomas’ NHS Foundation Trust and was outlined in chapter 2 section, 2.3.4, could help in addressing a broader system error of not prioritising and in some cases, avoiding these conversations. Back, Arnold, Baile, Tulsky and Fryer-Edwards (2005) researched effective communication skills in oncology specialists and they articulated: “There is a tendency among oncologists to respond to patient distress with more chemotherapy” (p. 175). These findings illuminate the broader scale of this problem in the clinical world. This is arguably more of a problem when discussing rare diseases due to the unpredictable and sometimes unforeseen basis of prognosis due to the variability of the conditions. For people living with NPC no specialist nursing support akin to Macmillan or other higher prevalence diseases is available at the time of diagnosis, leaving people potentially more vulnerable.

Some of the variation in communication by clinicians may be to do with feelings of guilt or failure surrounding their inability to cure NPC, alongside their recognition of the limits of what can be done. Medics have tended to objectify disease, seeing it from the perspective of normality, locating it outside the corporal space (Armstrong, 1987). As explored in chapter 6, section 6.1, the advances of medicine and development of hospitals have opened up the movement from caring for a person in a meaningful way to curing a patient from disease. In the case of rare disease, advances have not been as progressive but within the medicalised world, the non-treatment of a condition means failure and the role of the HCP is left void (Benson, Schwarz, Tofle, & Parker Oliver, 2018). This can also be viewed from Heidegger's (1977) essay on the spirit of technology where he suggests that technology is defining both the world in which we live and how we, as objects ourselves, relate to it (cited in Galvin & Todres, 2012). As we come to re-understand ourselves objectively in light of this, we run the risk of distorting our self-concept as a result of "our unspecialized capacity of being" (Galvin & Todres, 2012: p.38). Within the context of medical science, this leads to a more dehumanised approach to care as increasing levels of technology distance us from the humanity it is designed to serve.

A consequence of living with an incurable LLC that is rare and often unrecognised by HCPs was that people I interviewed described themselves as becoming the expert, communicating 'objective' information about the medical explanations of NPC to HCPs (Walseth & Schei, 2011). Although there is a beneficial dimension to this in terms of being able to understand in greater depth the disease manifestations and to be more informed as to appropriate care pathways, it emphasised the isolating nature of the illness experience exemplified in the lack of clinical support. This echoes Mishler's (1984) findings on the competing voices that are present in consultations, a dichotomy between *medicine* and the *lifeworld*. When the voice of medicine is the loudest, the result can be increasingly dehumanised care. This was found in other research where people living with a rare disease felt that they were obliged to provide diagnostic information to the HCPs that they came into contact with (Brodin, Sunnerhagen, Baghaei, & Törnbom, 2015; Budych et al., 2012). As Walseth and Schei (2011) argue, patients are in need of more than just disease-focused understanding, they also need "motivation and courage" from HCPs to contend with their illness experience (p.82).

In a study looking qualitatively at the experiences of parents navigating the healthcare system who had children diagnosed with a rare disease, findings showed that parents felt that their becoming an expert in their child's disease was not supported or acknowledged by HCPs in such a way as to encourage an egalitarian relationship (Baumbusch, Mayer, & Sloan-Yip, 2018). This denies the opportunity for the kind of participatory decision-making process that is advocated by research, whereby decisions are negotiated between the HCP and the patient; taking into account the desires and understandings that the patient holds (Horne, Weinman, Barber, Elliott, & Morgan, 2005). On a separate point, parents seeking this level of expertise in the illness in question may be understood as a type of coping mechanism, a 'performance' of knowledge in order to mask uncertainty and vulnerability (Heath et al., 2016). There were points in the narratives shown in the studies in this thesis where similar findings were present, and these seemed to be at crucial moments, such as when making end-of-life care decisions. When continuity of care was shown, this led to productive and effective clinical experiences, highlighting the importance of consistent HCPs and the meaningfulness of relationships. Caputo (2014) suggested that the relationship between people living with rare disease and HCPs should be handled with continuity, with HCPs providing medical care but also addressing the emotional concerns of living with disease. Evans and Rafi (2016) suggested that people with rare disease seek a collaborative approach that empowers the person and family impacted by the diagnosis, an experience that was not often found in the stories told by the families I interviewed. Dudding-Byth (2015) argued that people living with rare disease are the ‘expert patients’, thus challenging the traditional perception of the patient-doctor relationship. He encouraged clinicians to acknowledge the expertise of the person living with the rare condition alongside the person recognising the skills clinicians have, creating a partnership.

With only one specialist nurse in the UK for people living with NPC, more needs to be done to support this population in terms of clinical services offered. The lack of awareness of effective services for people living with rare disease has been acknowledged by the Department of Health (Dodge et al., 2011). In 2010, the UK’s chief medical officer, Liam Donaldson called for urgent measures to be taken to improve the awareness of rare diseases, saying that “children are dying needlessly due to misdiagnosed or undiagnosed conditions” and called for more funding for services in the UK to support the rare disease population (Dodge et al., 2011: 791). Although the Department of Health and Social Care (2018) suggested strategies are in place for people impacted by rare diseases, many of my participants were not involved in and/or did not know of any local, official routes of support such as support groups. Evidence has suggested that this is due to the lack of funding that goes into these groups in combination with the postcode lottery nature of it (Augustine, Adams, & Mink, 2013; Rodwell & Aymé, 2015). In addition to this, help from social services was a source of contention for quite a few of my participants, who described the convoluted process to secure equipment or financial benefits to which they were entitled. With this said, more than half of the participants in this study had been in contact with the NPUK charity in some capacity, thus were aware of a disease-specific hub, offering support.

A target more easily enacted would be to focus on the development of good communication skills amongst HCPs when giving diagnosis, prognosis and more information about the disease. Even if HCPs are unsure about this, listening to concerns, planning ahead of consultation and signposting in other clinical directions would help people in the initial stages of diagnosis. A beneficial model to consider in these situations may be Habermas' theory of communicative action, whereby rational conclusions of what is practical, right and good for the patient can be established through communicating at a lifeworld level. Habermas describes three different dimensions of the lifeworld: the objective, the social and the subjective. Walseth and Schei (2011) advocate that a medical conversation should include aspects of all of these dimensions. They suggest that for communicative rationality to occur, decisions have to be rooted through "lifeworld mapping", thus strengthening the relationship between patient and HCP (p.88). This complements findings from Dahlberg, Todres and Galvin (2009) advocating a lifeworld-led care as opposed to a person-led/patient-centred care approach. This demonstrates the importance of agency and vulnerability from all parties, as opposed to the focus of much patient-led care, which has mainly been translated into recommendations for practice that includes "focus on systems" and "conditions of service" (p.226). Instead, lifeworld-led care seeks to use the elements of the lifeworld to "form the parameters" from which individuals and families experiences could be understood, and how these descriptions could be central in leading humanised care (Dahlberg et al., 2009: p.266; Galvin et al., 2018).

Temel et al. (2017) suggests that when people are diagnosed with a LLC, the option of palliative care should be spoken about at diagnosis to encourage people to live well with deteriorating health. As seen in chapter 2 there are mixed opinions as to when palliative care should be discussed, with some suggesting action plans for end-of-life care being brought into the last few years of life as introduced in the advanced care planning gold standard (The Gold standard framework, 2018) and others suggesting in the last year of life (Kimbell, Murray, Macpherson, & Boyd, 2016). This uncertainty is difficult to judge given the unpredictability of the prognosis in rare diseases and the likelihood that such conversations are held at crisis points for the patient and their family. These crisis points are likely to be due to resistance to conducting conversations prematurely (Mitchell & Dale, 2015), or the lack of communication from HCPs in supporting people in the transition to palliative care services (Abarshi et al., 2011; Newbould et al., 2012). This variation of thought illuminates the lack of consistency of care, where people could be missing out. The safety of the hospice was described by Jeremy in chapter 6, section 6.2.5 as an embrace, and many of the participants that I have interviewed described the positive experiences of day/weekend visits to hospices, a trend supported by other research (Gawande, 2010). Anecdotally, it took some participants a while to start attending the hospice, due to the connotations that the hospice setting holds as a place designed for death. In addition, perceptions of 'the hospice space' seemed to confirm the realities of the dying body, and sometimes this was avoided in a way that corroborates the trend of avoiding death observed more widely in society (Mannix, 2017). The taboo of death has defined society so profoundly that we resist talking about it in case it kills us (Kortes-Miller, Boulé, Wilson, & Stinchcombe, 2018).

It can therefore be noted that the overall clinical experience of those living with a rare disease can be characterised through a relationship to knowledge, or its absence, in the individuals themselves, their carers and the HCPs they encounter. All stages of the illness experience emphasised the interface between knowledge, perspective and communication.

8.2.2 Discussion of the impact of illness on the Lifeworld

This shift into a world of illness was shown to have fundamentally altered how people impacted by NPC experience and understand the world around them. Svenaeus (2011) argued, in illness, “the body shows up as an alien being (being me, yet not me) and this obstruction attunes the entire being-in-the-world of the ill person in an unhomelike way” (p. 337). The findings of this thesis regarding the ways in which the elements of the lifeworld were affected in people impacted by NPC will now be explored.

Temporality

The concept of time was a powerful element in this study. The differing ways that temporality was experienced throughout the lifespan demonstrated the different needs people have. For young children, a sense of the fragility of time was not shown to be something that had been contemplated in conscious thought. Expectations for the future were held; there were still occasions to look forward to. The subtle expressions in the temporal were felt when looking back on the capabilities of what could be done in the past compared to what they could do now. An experience analogous to ‘Benjamin Button’[2] was conveyed, where the temporal and developmental milestones were inverted, leading to a disrupted and discomfiting narrative characterised by loss and alienation. For parents of children with NPC, this was experienced at its most potent in those who attended the annual NPUK conferences; an environment where they were faced with mortality and death year on year. A future orientation meant more deterioration, more incapacity compared to what was. Death was something that was not spoken about as being a concern to children and this may be due to their limited conception of its realities, supporting the idea that with age comes a more nuanced and realist understanding of death (Bonoti, Leondari, & Mastora, 2013; Speece & Brent, 1984).

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[2] Referring to the film, ‘The Curious Case of Benjamin Button’ (2008) where Benjamin Button is born as an elderly man and ages in reverse.

For adults, time could be described as being ‘dislocated.’ The initial ‘narrative wreckage’ (Frank, 1995) of receiving a life limiting and incurable diagnosis was evident throughout this study. The past echoed opportunity, as lives were continued to be built. The initial reflection on the permanency that an incurable, progressive illness caused meant that both the present and future felt hopeless. A land of limbo was portrayed, with a felt sense of there being no way forward, a sense of entrapment (Gendlin, 1981). For some, as time progressed a wavering acceptance was found, which enabled the present to be something that had a sense of mobility. For others, a present sense of mobility was yet to be. The future that was met with trepidation, when close to death, time was juxtaposed with mortality. On one hand, time was fleeting, as the acknowledgement of loss abounded. On the other, the passage of time slowed, especially when both objective and subjective suffering was present. Time had a different meaning at the point of death. Past worries and struggles were not thought about. Time was precious in the present, using every opportunity to be with the dying one (Kastbom, Milberg, & Karlsson, 2017). The results illuminate the vulnerabilities of time for people with NPC, which from my understanding seem to be novel findings compared to other research looking at rare or LLCs. The eclectic experiences of time should be recognised, with greater support being given to helping people feel at-peace in the present, in order that they might better conceptualise their future.

Embodiment

The objective body of those living with NPC progressively deteriorates and the body becomes unable to work. There is a relationship to be explored here between the objective functioning of the body and the subjective experiencing of the self; experience shaped by one’s ability to move, eat and think (Finlay, 2003). The disease causes these functional breakdowns but the physical manifestations of these are outweighed by the impact they have on the ‘self’, whose life narrative is permanently changed as a result. It went deeper than learning that they had a disease and instead transformed their whole meaning of self (Finlay, 2003).

For children living with NPC, aspects of the bodily-self feeling low, depressed and confused were found as their objective body reacted to NPC. How the body mutually interacts with the world was shown in an *in-out* notion, the immobility of the present compared to their past ability, leading to facial manifestations of depression, thus impacting upon their mood (Todres, Galvin & Dahlberg, 2007). The bodily self had to realign with the boundaries that the objective body had enforced upon it, such as equipment now being used to help the body position itself in a desired space. The relationships between the bodily self of children with NPC and their families were seen as important through the restoration of dependence in an unnatural way, against the grain of becoming independent.

The lived experience of NPC for adults was shown to fundamentally alter their relationships with their self and with their body. A disconnection from the body once known had occurred (Charmaz, 1995). This disconnect has been presented as *bodily alienation* in other phenomenological illness explorations (see Charmaz, 1995; Finlay, 2003; Svenaeus, 2011). The body had become more known, as opposed to taken-for-granted. This went deeper than the mechanic, cellular level, transcending the sense of self in such a way that was dependent on the occurrence of illness (Charmaz, 1995). The union of the objective and subjective was seen in the illness experiences. This was seen in language used, in metaphors about the isolation of the illness experience, in distress about the future and the continuation of the loss of the self, in the working out of how to live with this new sense of self. This manifested in many emotions: anger, impatience and depression. A letting-go of previous expectations and a learning of what would now be was a continual process as the experiences and emotions of disease manifested causing deterioration. Carel (2013) spoke openly about her illness journey and how her body formed new habits in negotiating the world, describing "blissful forgetfulness of the pleasures of physical movement" (p.30). In this study, some participants were still stuck in this change of self-body-world shock, showing the 'realisation' of the changes of the body (Finlay, 2003). Others were re-learning how to manage the continual process as the progression of the disease took over the body more and more, each time adjusting the intertwined self-body-world horizons.

As death was pressed upon, there was a sense of acceptance, a surrendering to the fate that was known all along. No more can be done. Though the body dies, the sense of self being meaningfully situated in the world lives on after death, through the fundamental relationships with the interpersonal world that they were once a part of. This finding complements research that has argued that the embodied relationality that illuminates the connectedness that is inherently experienced, continues after death in material practices and felt experiences (McCarthy & Prokhovnik, 2014). This is shown in this study by the retention of ashes on the bedside table to maintain the relationship with the dead. Other studies have shown a visible form of embodied relationality in the form of ashes made into jewelry (Prendergast, Hockey, & Kellaher, 2006). For those in the present study, death and bereavement was an imposing and definitive moment, but it was not an end; something of an essence continued, as did their story. The findings in this section draw on the work of Merleau-Ponty (1962) in recognising that our embodied experiences cannot be separated from our sense of self and our being-in-the-world. The relationship between body and world is wrapped within "flesh of the world and myself as flesh" (Aanstoos, 1991, as cited in Finlay, 2003: p.158) intrinsically related to others and our position in the world.

Intersubjectivity

This present study found that the way that people with NPC and their carers see themselves, and their own illness disruptions were through the eyes of the other. This restricted people in their sense of freedom, in what they could not do compared to the other. This drove decisions to retreat from the world around them, where NPC was unheard and unrecognised. Parents who were caring for their son or daughter with NPC became closer, relying on their shared experience with each other. Children with NPC were shown to value the family environment and their siblings, offering a focus and distraction within their illness experience.

For adults with NPC there seemed to be a mutual separation between friends; friends were perceived to not understand NPC and people with NPC perceived their friends to be moving on with their own lives, where there was no room for illness. As Carel states: "Illness is a theme that can envelop one's whole life. But it can also – and often does – recede into the background in a way unimaginable to the healthy outsider" (Carel, 2013: p.244; Strauss, 1975, cited in Bury, 1982).

The findings of the studies in this thesis demonstrated the sense of loneliness that permeated in the illness experience causing years of depression for some, yet this was preferential compared to seeing the self through the other. The existential assumptions that some participants had in relation to others were threatened by illness as their world became unmade, showing the inherent intertwining of body-self-world being one. Caswell and O'Connor (2015) argued that some people exhibit agency in withdrawing themselves from society when terminally ill, entering a self-imposed period of social death, where they cease to participate in the social world, prior to their biological deaths. Across the studies in this thesis a similar retreat from society was found. Yet, reliance on families was a key finding thus not resulting in complete isolation. This was especially shown in chapter 6, in dying, thus choosing to experience this time in the intimacy of the nuclear family.

Yet, with this said, for a lot of people in this study, the NPUK charity was a great source of support where understanding was found. The shared, insider experience of the disease and the complexity of the illness experience was a platform where suffering could be understood. At the annual NPUK conference, meaningful discourse, which resonated, created a sense of belonging, an awakening that they were not alone. The importance of having an open space to share narratives about suffering within the illness experience was found. Having the opportunity to do this created a sense of movement, a sense of at-homeness within the illness experience; a sense of becoming one with the situation, the body that is now being experienced within the world. They were able to find a location within the world, with people who had a more subjective understanding of their lived experience. It helped people to rationalise their position and offered a sense of humanising relatability. For some this created a mobilisation on leaving the conference towards a connectedness and ability, especially for carers to continue in their care. The importance of psychosocial support in meaningful shared experiences is seen in other rare disease groups (Anderson, Elliott, & Zurynski, 2013). Yet, quite often, the internet becomes the most accessible form of support when reading about others' experiences of living with rare disease, relating to others from afar (Garau, 2016).

For others, the conference community did not offer the depth of attunement that they needed. The heterogeneity of NPC and indeed illness experience meant that the experiences were so varied that they felt they could not relate to others in a meaningful way. This highlights the idiosyncracy of narratives of illness experience emphasising that care should be tailored to the person and not the disease.

Spatiality

When referring to spatiality it must be understood as both a physical and ontological concept; it is as much to do with one's sense of position in the world, one's being, as one's physical presence. It includes the space we inhabit, but also the things and people we experience the space with and through. The notion of space was also relevant in the sense that the reality of living with NPC was "close" in everyday life, as other materials faded into insignificance. For some the environing world shrunk as diagnosis was given and the illness experience progressed. Feelings of imprisonment and exile from the world that was known impacted upon mood. Hospitals often were represented as: a factory where intervention was provided to solve a physical problem and once 'solved' they would move onto the next patient; an undesirable place where painful procedures were conducted and artificial sense of caring space was felt in the midst of bleeping machines and strangers; a space which felt hostile. There was also a perceived failure in the sense of the hospital being a safe place due to the lack of understanding of the disease. In chapter 6 we see Philip's mother choosing to remain a constant presence because she believed she knew more about how to care for him than the nurses.

Outside of the hospital setting, comfort was found at home and in the openness of their gardens. The sense of the environment and the landscape with which we are surrounded goes beyond the notion of an attractive view, it speaks to how an individual experiences the world, beauty and peace are embodied in our experience of them, they become an integral part of our interpreting of the world and our circumstances (Brook, 2013). The architectural design of the hospice spoken of in chapter 6 brought comfort and openness at the end of life, contributing to an abiding expanse, *Gegnet,* to one of the most difficult times one will experience. This served to highlight the importance of carefully considered palliative care. Public policy within palliative care has concentrated on granting the preferential place of death, but the main place of death is still the hospital and thus the spatial elements which impact upon feeling at-home whilst dying are not being met (Oxenham, Finucane, Arnold, & Russell, 2013). In 2016, almost half of all deaths in England occurred in hospital and less than 6% of deaths were in a hospice (Public Health England, 2018). Although preference of dying at home may be conversed, this requires complex factors both in terms of community health services support and informal networks of care to offer help (Horsfall, Leonard, Rosenberg, & Noonan, 2017). This in turn with the medicalisation of the era means that the importance of spatiality in death is difficult to take on board. Thus the majority of the population is not experiencing an environment that meets their needs and desires at this key point in life. As seen in chapter 6, those that did experience the space of death they desired, gained in elements of existential well-being during the dying experience and afterwards.

The intricacies of the meanings of space were present in offering comfort or discomfort, a sense of openness or of imprisonment, freedom or entrapment. Space can provide opportunities such as the awe felt looking at nature that helps when grieving, or the closing of horizons and the sense of imprisonment when feeling unsupported by social services. These examples serve to demonstrate that space represented an important framework and filter for the illness experience.

Identity

As discussed above in intersubjectivity, the interpersonal relationships and interactions with society were impacted upon when living with NPC. This was a contributing element interweaved within the change that one's identity went through. This in many contexts affected the self-growth of a person, where isolation from the world around them was something enforced upon them (Charmaz, 1995). The fundamental identity of children was changed when they could no longer play with their toys or take part in activities such as dancing as seen in chapter 4. There developed a greater emphasis on watching their siblings or family members to seek pleasure and reassurance from them in some vicarious sense. This movement in identity became more of a reliance on known others, on family, in comparison to when they were more able.

Some of the adults in this study were seen to become more hostile to others; their presence and voice became more silent. The links with others became looser and interactions were not mutually appreciated as they had been in the past, as illness changed the objective body.

Adults were found to pose themselves as the problem, as the one who is ill and no longer able. Perceptions were that others saw them as illness, as becoming increasingly unable, which shifted them away from previous mutual appreciation. Aspirations shrunk along with a sense of their identity, their capabilities. As Charmaz (1995) found: "preferred identities that people assume, desire, hope or plan for" (p.659) were beaten, which impacted upon their mobilisation, of moving forwards with life. For some, a sense of agency was re-established and a sense of movement was more apparent than for others, where a re-learning and establishing who they now were was taking place.

For parents of sons and daughters with NPC or for those whose spouse had NPC, their identity had changed to becoming a 'carer'. A poem written by one of my participants conveys this felt change in their identity:

The Prisoner

From my childhood
I remember a scene in a history book.
A man in chains sits in the centre of a cold flagged floor,
and as if on stage,
is encircled by a pool of light,
which streams into the gloomy cell from a small window, high above.
The thin, tapering cone of brilliant light
is all that links him with the outside world.
Its soothing warmth thrusts aside the cold darkness of the room
and reminds him of the other world that once was his;
a world of freedom, enjoyment and friendship.

But now I am there, for I am that person in the room,
imprisoned not because I have erred,
but because I choose to care.
The picture in my mind is now my reality:
For I am a carer.

The sacrifice of caring for their loved one is shown here. The juxtaposition of the admirable qualities of caring with the claustrophobic and punitive imagery speaks to the internal conflict present in the lifeworld of those whose sense of self is disrupted by childhood memories. The temporal dimension is present in the reference to childhood, a time of hope and disconnection from the suffering being read about. Illness has brought suffering, it has made it real and moved the notion from the theoretical to the experiential. The physical space described is indicative of an existential reality, one that speaks to the embodied sense of oppression and burden. One must also note the sense of injustice present in the language, a sense of one's identity being unjustly derailed by one's own virtue, the writer is wronged by circumstance, and then this is exacerbated by the decision to care. Caring brings a burden, caring removes choice and self-determination, caring defines your whole being.

The intricate links identity has between embodiment and intersubjectivity are seen in this poem. The embodied self questions his identity and relations with others. Yet movement was found in stepping into the new identity, the new sense of self that is found in the world of illness, where compromises are made.

Mood

Enwrapped within each element is the powerful dimension of mood. Mood shapes the elements and is complex.

> "[Mood] is not just an internal happening, but is perceptual and interactive… so in both illness and well-being, the description of mood as a qualitative dimension is part of a holistic, coherent understanding." (Todres, Galvin & Dahlberg, 2007: p.58).

The complex and intricate experiences of mood were unveiled, wrapped within the elements previously spoken about. Mood was seen to create a sense of flow forward towards an at-homeness *or* backwards, generating a sense of a lack of equilibrium, attunement with anxiety and of disturbance being inescapable. A sense of either openness or 'closed in' mood either created light on the horizons, offering mobilisation, or darkened the horizon, with a foreboding inability to continue. The influential element of mood can shape people in how they respond to a diagnosis, live with NPC or care for someone with NPC (Todres, Galvin & Dahlberg, 2007). The ever-changing nature of mood further complicates the illness experience, with the presence of well-being, of authentic living, of acceptance being intrinsically wrapped within the depths of suffering and immobilisation. This well-being-suffering co-existence, will be further explored in the next section.

Summary

The experiences of people with NPC along with their carers in this programme of work has illustrated the profound impact of illness on the lifeworld. The elements of the lifeworld elucidate the existential impact of NPC as illness disrupts the individual's narrative. The interpenetrated elements have outlined an exploration of how one lives with and cares for someone with NPC and the impact this has on a person, far outweighing a pure focus on the cellular or physiological level of disease. The central findings focus on embodiment, learning how to live with disease and the close links it has with time, space and relationships; echoing that the lifeworld is therefore not about inner experience but outworked in intricate and outwardly visible ways (Finlay, 2003).

8.2.3 The co-existence of suffering and well-being

Illness has sometimes been described as something that it is possible to enjoy, where wellness can still be experienced (Carel, 2017; Broyard, 1992 cited in Frank, 2007). Thus, the use of the word 'disruption' in this thesis may not be favoured by some due to the negative connotations that this suggests to people who are living with illness. Yet to take 'suffering' and 'illness', 'well-being' and 'health' to be close cognates does a disservice to the complexity of the concepts (Galvin & Todres, 2011). By engaging with both well-being and suffering, the findings from the thesis have shown the intricate ways in which people living with NPC experience both, in complex and ultra-fluid ways. I will now move on to explore these existential encounters and postulate the sense of at-homeness that was found in these families.

The essence of well-being as being-at-home which offers ontological security in relation to the self and others was found in this programme of work (Giddens, 1990). It is the co-existence of dwelling (peace in the moment) and mobility (opportunities for movement) that creates the best possible sense of well-being or the worst possible sense of suffering – *Gegnet* (Galvin & Todres, 2010). It is in this way that a sense of at-homeness is fully understood. Another conceptualisaion of at-homess is offered by Seamon (1979) who suggested five pre-requisites that contribute to a sense of at-homeness, these are:

- Rootedness (a sense of familiarity and belonging)
- Regeneration (the restorative function of place)
- At-easeness (the freedom to be oneself)
- Warmth (a friendly and supportive atmosphere)

- Appropriation (a sense of possession and control)

Seamon's (1979) work resonates with Galvin and Todres' (2010) conceptualisation of at-homeness. Aspects from both of these attribututions of being-at-home were found in my participants, especially for children with NPC. This was mainly found in regeneration and rootedness in place and belonging. The security of home and the connectedness to close family held relevance and created a settledness, showing how experience is co-created by the individual and their world (Giorgi & Giorgi, 2003). With relation to well-being, emphasis here was seen more on the security of dwelling rather than the expectation of future possibilities. The fragility of life and what it held for children living with NPC created a driving force for parents to make sure that their children, along with themselves, enjoyed the present. The pursuit of this was evidenced in the organising of significant holidays to Disney, days out or similar. Illness had provided a greater appreciation of the present, a sense of privileging the present in a way they had not before, offering a regenerative experience, aligning with the idea of the authentic life (Carel, 2013; Heidegger, 1962). The future brought with it deterioration and inability to do the things that can be done in the now. A distancing of self from the future and even from the past in what was possible then, provided a way of enjoying the now, finding a present-centredness in dwelling. An Epicurean approach to life was shown, concentrating purely on living in the present, in the moment, recognising that its joy will result in freeing oneself of the constraints that the past and future hold, thus achieving well-being (Carel, 2013). However, a Heideggerian approach to authentic life is one where we live in a way that acknowledges death and thus develop a mindset of being-towards-death. This does not shutout the future, but recognises it in spite of what may come.

The lived experience of time enwrapped within the sense of self and objective body of adults living with NPC was something which caused a sense of homelessness. This may be due to the adults in this study being diagnosed in adulthood, thus, remembering the life once lived in a more vivid way and the drastic changes that illness brought to bear in comparison to the children in this study. Similarly, it should be noted that the nature of parenting means taking children under the wing, which has significant implications for parents' perceptions of illness. For adults, greater recognition of the nature of life and death led to discomfort, unease and anxiety. This was accompanied by the sense of moving out of control of circumstance, a threat to their own self-determination. The threat of progressive deterioration placed their goals and dreams at risk, exposing their fragility. A preoccupation with the present became the way to live, as a force to make the most of the time they have left. This has been found in another study looking at the progressive disease Parkinson's, with a carpe diem attitude being shown, a desire to making the most of the situation (Smith & Shaw, 2016). Yet, this would suggest a freedom in movement, to seize the moment. However, the results in chapter 5 increasingly unveiled a sense of entrapment, which triggered a disengagement; a dwelling, which did not have at-home-like qualities, but instead felt uncomfortable in the extreme; the essence of suffering.

The longstanding impact of diagnosis on the lifeworld was presented. This was characterised by the difficulty of generating a sense of momentum following diagnosis with NPC, a sense in which the self would be able to move on from the present crisis. There was a need for acceptance of the reality of life with NPC, a need to recalibrate before being able to move forward. This was not in the sense of fighting against their diagnosis, but rather an overall acceptance that they had surrendered to defeat by NPC (Charmaz, 1995). This could be understood as a paralysis of living in the present, a non-connection to their body, a foreign body preventing mobility towards a future sense of at-homeness. An example of the sense in which acceptance and surrender co-exist is found in Charmaz's (1995) study of adults with serious, chronic illnesses that: "The quest for control over illness ceases and the flow with the bodily experience increases. Surrender means awareness of one's ill body and a willingness and relief to flow with it" (p.672). Some had tried to step into this 'new world' of learning how to exist, but an oscillation across a spectrum of experience was portrayed, in the intricacies of possibility and suffering. Yet learning of a new way of being-in-the-world was presented, as Merleau-Ponty labeled, "Restrictive potentialities" (1962, p. 143) wrapped within the midst of a resignation to the fact that improvement will not come (Charmaz, 1995).

Suffering was not necessarily seen in the pre-reflective, objective bodily deterioration. As found in this thesis, pain is not an essential part of the suffering experience and instead can be embodied by characteristics of loneliness, fear, loss and a sense of hopelessness (Reed, 2003). This can lead to lack of autonomy and the loss of individual's humanity (Rodgers & Cowles, 1997). The discourse of 'suffering in silence' highlighted the chosen isolation that some suffering can take place in, as relations break down, especially for adults living with NPC. The bodily experience of suffering was found in the alienation of seeking belonging in a 'healthy world', thus a loss of attunement was seen with one's sense of self, one's relations and one's position in the world causing a disharmony in the present which shaped the future.

Overall, shared experience of suffering across the lifespan of living with NPC was shown in the combination with the obstruction that illness had caused in the body and the sense of being-in-the-world in an unhomelike way. The journey of finding a sense of at-homeness was something that was found in the levels of suffering and well-being spoken about above. Thus the illness experience was not something that solely determined suffering or well-being, but rather the levels were existentially lived. As a result, living with NPC was shown to impact upon the meaning-making process of being-in-the-world. As Sontag (1983) writes: "we are each citizens of two kingdoms, the kingdom of well and that of sick" (p.3). For those who are living with an incurable illness, the kingdom of the sick may now be a permanent residency. The epistemological heritage of the language of two kingdoms can be found in early Christian theology: the Augustinian notion of the saved and the damned, the chosen and the rejected, the hopeful and the hopeless. To explore and critique this notion further, one can look to the development within such thought of a purgatorial state, a border between heaven and hell where souls lie awaiting judgment; a place of limbo. This concept of limbo is explored in Levine's (2013) anthology surrounding stories of caregiving in people who are terminally ill and is here described as a state of doubt combined with feelings of being trapped, whilst waiting for events beyond one's control to unfold. If one has a foot in both camps, if one travels between the two, then one travels between a sense of at-home and home*less*ness. For those in the midst of the illness experience, those lost in purgatory of suffering *and* wellbeing, finding a sense of home is elusive and evasive, found fleetingly and greatly prized. This is an example of the *Gegnet* of suffering, to continue the quest to identify how that state of being might come about and what its experiential quality might be like.

Homecoming

Were experiences of homecoming re-told in this programme of work? I would argue that aspects of at-homeness were experienced by my participants, yet on the whole, the process of re-adjusting to life with NPC had not quite been fully realised. Heidegger (1962) suggested that to live an authentic life, an individual must grasp the inevitability of their own death. Thus in and through suffering, a more authentic appreciation of life may be had. This speaks to a paradox at the heart of the relationship between suffering and well-being, in that it is in the midst of the anxiety and suffering of homelessness that one can find the motivation to seek a sense of homecoming. I would argue that the most meaningful lived experience for an understanding of well-being and homecoming was found in the face of death for the two families whose children had died (chapter 6). A sense of the authenticity was presented, in that they had undergone the suffering, and the greatest of losses, but through this could lead a more authentic life than they previously experienced. This emphasises the elusiveness of well-being in the context of illness. It can be found but cannot be assumed; it requires careful cultivation, courage and the willingness to change. This programme of work elaborated on the individual meaning of these experiences and the journey that brought them there. The adaption of each individual to the body as foreign or alien was the point at which these stories reached their common conclusion, the point which these stories were left to continue unobserved. Each journeying towards the person they were becoming, 'the next viable them' in a continuum of self-reconstruction and renewal.

8.3 Moving forwards

8.3.1 Giving voice to the journey towards homecoming

The main body of findings in this programme of work suggest firstly that people impacted by NPC experience a permanent disruption to the life they once knew; a bodily experience that impacts on their self, time, space and relationships. A co-existence between the existential experience of well-being and suffering occurred. I would argue that this existential experience deserves a voice. This section will focus on the possible ways in which a sense of at-homeness for people living with NPC may be supported; well-being as the goal for people living with NPC that is much deeper and more complex than the absence of illness. I agree with Frank (2013) in that one of the main ways that this can be done is through the shared experiences, the shared voices of living with NPC and more generally, rare disease, in the form of stories. The emphasis on shared insight and articulations on shared experience of suffering could assist others in their thoughts of becoming aware of their own. Frank attests that, "because the pedagogy of suffering is taught in the testimony of illness stories, the kind of ethic it supports is a narrative ethic" (Frank, 2013: p.154).

Stories were told in chapter 6, where the direct experience of NPC had come to an end. Through the death of their loved ones, they continued to tell their story of suffering, of loss of self. Yet, their new understanding of anxiety gave them a fresh understanding of what being-at-home means. Both families felt the sense of responsibility to guide others in the same position. Bearing witness to their life with illness represents an experience beyond the explanatory power of medicine alone. The legitimacy of the experience of the individual can and should contribute to society's understanding of illness and well-being. Frank (2013) spoke openly about a challenge that he found when he was living with cancer. He articulated in his preface: "suffering does not magically disappear when the tale is told, but the more stories I heard the less space my own suffering seemed to take up. I felt less alone."(p.xi). This could arguably be even more necessary in people living with rare disease, in making people aware of what living with illness is. Even though the illness experience may vary on multiple levels, according to culture, context and across time, shared appreciation of this can encourage empathic care that is more humanised (Galvin et al., 2018).

The act of story telling can be therapeutic, reaffirming the experience and perceiving the journey they have been on (Greenhalgh & Hurwitz, 1999). After I had conducted my second interview with the Bentley family in chapter 6, they thanked me for conducting the interview and for my interest in their story. It was my privilege, but it spoke about the power that speaking out your story has, the aesthetic of life, both in terms of well-being and suffering. It also spoke of how this is not done often enough; stories are left on the shelf. The shared experience, the shared heritage, provides common ground for shared humanity and is enabled by our sharing of stories, thus potentially supporting people in stepping towards living more authentically. Transferring a storytelling movement into the clinical world would give clinicians a real understanding of the multifaceted aspects of living with illness in order to suitably support people, something that was missing in the experiences conveyed by my participants.

“Listening is hard, but it is also a fundamental moral act; to realize the best potential in postmodern times requires an ethics of listening” (Frank, 1995: p.25).

8.3.2 Quality of life scales

The tragedy alongside the new insight of meaningful life that has come from living in a world that has been wounded by an incurable illness has led me to scrutinise the meanings, both implicit and explicit, of the QoL scales that I have developed and whether these resonate with people in this position. With this in mind, I think the findings of this study warrant a careful consideration of the appropriate use of the term QoL within questionnaires for people living with a LLC. The existential understanding of well-being surpasses what happiness means and instead is a deeply embedded way of living which recognises and incorporates anxiety in a Heideggerian sense that leads to an authentic existence. This is not to say that traditional QoL scales do not meet with existential issues; rather, I think the findings of this study support the idea that people are not initially seeking to enhance their life but instead accept the life they have in a way that is authentic and thus acknowledge the intrinsic connections between well-being and suffering.

With this said, throughout this PhD experience, I have come to realise that what I think is lacking in the clinical world that people with NPC often are involved with, is a tool that helps them convey their illness experience to HCPs in such a way that engenders meaningful dialogue. It is necessary to create the opportunity for dialogue that is lifeworld-led and two-way, in a way that engenders trust that will cushion decisions that need to be made and help people to feel more supported (Dahlberg et al., 2009; Walseth & Schei, 2011). Borg Xuereb, Shaw and Lane's (2016) paper exploring consultations between patients and consultants found the ideal health encounter to be: “an open space where patients are offered accessible information and time for consideration; an opportunity for physicians to determine what matters to patients within their life context when making a treatment decision; a collaborative consideration of options taking into account the evidence base and what is right for the patient; and a two-way dialogue where physicians adopt academic humility and patients feel genuinely confident to make an informed decision that is right for them." (p.449).

On a pragmatic level, the findings in this study lead us to a point where the intersection between the lifeworlds of people living with illness and clinicians need to come together. This is similar to Gadamer (1975) notion of horizons in the sense in which our worldview and belief system interact with those of others. The need for these horizons to overlap is a prerequisite of effective communication (Shaw, 2010) and the fusion of horizons is made possible when we are transparent and vulnerable. Within the healthcare setting, this would take seriously the arguments of Toombs (1993) that clinicians and 'patients' live in different lifeworlds, with different core beliefs about meaning, significance and relevance. Both parties need to be heard without succumbing to claims of aloofness or noncompliance. This is necessary for the evolution and development of the care that hospitals provide for people, particularly those with rare diseases. Such practice would have to reckon with the fact that the stories recounted by patients when talking about their experiences, often lack a clear order and sequence that would make them more readily understandable to a clinical setting defined by structure. This requires a different way of being as a HCP, requiring training akin to that of person-centred therapy (Pryce et al., 2018). This is compatible with Elwyn, Edwards and Britten's (2003) definition and use of 'concordance' to describe the relationship between HCP and patient; replacing terms that have connotations of paternalism and authority such as 'compliance' and 'adherence'.

Using the questionnaires developed in this study as a tool for dialogue between HCPs and people with NPC is something that should be further studied. Items that resonate with people could be explored in greater fullness, discussed in conjunction with a deeper appreciation of the story being lived out. The scales contain items that are based on shared experiences of living with NPC, in a way that looks existentially at the meanings conveyed by people in this position. This of course is not an exhaustive list of items, to provide such would go against the principles of phenomenology. Yet, this could act as a guide for HCPs to firstly understand more about the deeply embedded humanly experienced nature of living with illness, and thus engaging more with humanised care (Galvin et al., 2018).

8.4 Contributions to knowledge

As the first qualitative study to explore the experiences of people living with NPC in the UK, this research makes a number of contributions to knowledge, especially in the areas of research development and healthcare practice, where insufficient knowledge of the experiences of rare disease is often the case (Pelentsov et al., 2015). This section will outline the contributions of this thesis, firstly through the demonstration that the substantive content of the findings is closely unified by the methodology adopted. In turn, the final implications for clinical practice and policy will be outlined, offering recommendations based on the findings and what future research should consider. The strengths and limitations of this programme of work followed by a reflexive account will conclude this chapter.

The research presented in this thesis makes a substantial and original contribution to research within the rare disease group, which is often undervalued but in need of support in order to understand more about the illness experience. It offers an in-depth analysis into the experience of living with NPC and has explored the existential nature of the ways in which illness disrupts the lifeworld. Not only this, but it has presented NPC from different positions of the life cycle, including the dying experience. Chapter 6 has also contributed to the existential work on suffering, death and dying research and suggested the importance of space and embodied relationality. The meaning of the aesthetic experience for both the person dying and their loved ones has shown that a sense of at-homeness can come both during that experience and afterwards. The rich descriptions of the illness experience of living with NPC are commensurate with Galvin and Todres' (2011) dwelling-mobility framework. Having this as this study's theoretical underpinning has achieved a different model to the more popular models of stress and coping frameworks such as Lazarus and Folkman (1984) and instead has offered a more open and nuanced basis of understanding the complex existential experiences of illness in a way that is sensitive to the need for humanised care (Galvin et al., 2018). This study has begun to gather evidence for the dwelling-mobility framework, appreciating the levels of well-being and especially suffering experienced by the participants. This body of work has also shown that through illness and death, people living with NPC and those who are caring for them might find authentic life, especially in dwelling. The findings suggest that the notions attached to 'mobility' are difficult to live with. This is likely to be due to NPC being an incurable illness, which therefore underlines the irrevocable changes to a person's life.

Another contribution this thesis has made is towards the conceptualisation of QoL. Moving on from more traditional understandings of QoL, and instead drawing on the lifeworld theory has provided a desired theoretical underpinning (Cohen & Biesecker, 2010), which to my knowledge has never been used before to develop a QoL scale. An idiographic approach meant a focus on the person's lifeworld as opposed to generalised invariant structures of experience, and offered a novel and in-depth exploration, alongside an examination of the meanings attributed to experiences. This was especially valuable when working with such a rare condition because it means that these meaningful experiences, rather than preconceived notions or concepts from other QoL scales, are used as the starting point for a measurement. Thus, the developmental stage of the scales in this study went deeper than other disease-specific scales, using an existential-phenomenological theory, which transcends the individual through drawing on existential dimensions of being human. The conceptualisation of QoL is therefore a more reflective questionnaire, which in turn allows for concrete experiences to come to the fore rather than relying on what might be constructed as more abstract concepts from other QoL measures for other conditions. Purely focusing on the recognised traditional structures and global themes of QoL may have presented a static picture, whereas, this analysis has elucidated the existential elements of a person's lifeworld (Frank, 2013).

A suggestion made by Råholm, Arman and Rehnsfeldt (2008) is for research to concentrate on understanding from an ontological perspective toward the world of illness that people live in, to present the unique sense of suffering in a meaningful way. I believe that the studies in this thesis have achieved this. This work can highlight to HCPs and researchers the individual experiences which may not always be visible in terms of the daily struggles and existential sufferings that families are living with within managing and negotiating relationships and their sense of self within a world that does not understand NPC (Petersen, 2006).

8.4.1 The methodological contribution

As discussed at the beginning of this chapter, I wanted to honour the stories that people shared with me and using a combination of methods helped to garner the experiences of living with NPC in a way that acknowledged the nuanced mixture of complexities and beauty. The utilisation of a phenomenological framework captured the richness of lived experience through exploration of daily life and has allowed us to glimpse the world of people impacted by NPC, which has not been done before. The employment of Galvin and Todres' (2011) existential-phenomenological theories of well-being and suffering guided me in being more open to unexpected discoveries when analysing these stories. The multi-dimensional levels of well-being and suffering widened the possibilities in understanding the experiences of living with NPC. Overall, adopting an explanation driven logic of inquiry (Hiles, 2012; Shaw et al., 2018), which draws on theoretical evidence and conceptualisation developed through other psychological and philosophical work, driven by the inductive method and abductive reasoning, allowed for a fuller picture of the illness experience giving the best possible explanation of results for this time. This was useful in seeing what my findings share with other experiences of illness, but also seeing how they differ from these studies, revealing new meanings and enhancing knowledge.

More specifically, adopting these methods meant that the meanings of living with NPC that may be imprisoned ordinarily were opened up to more expressive language and communication. New levels of meaning were especially found for children living with rare disease. The hermeneutic arm of phenomenology allowed an exploration of how parents make sense of their children living with NPC and presented an insightful use of discourse in exploring the lifeworld of their children. These approaches have led to producing ethically sound empirical research, issuing a deeper appreciation of the meanings of living with and dying from NPC.

As I progressed as a researcher over the course of this work, I sought to be more creative with methods. Seeing the value of this especially within the context of death and dying, I became acquainted with embodied interpretation. Inspired by Finlay (2006) and Todres and Galvin (2008) to not just recount stories, great freedom was found in being in touch with the felt-sense of my experiences meeting at the intersection between self and others. This encouraged me to seek the shared horizons I had with my participants, especially in terms of caregiving for loved ones. "Meaning and understanding do not rest only on thinking and watching, but also on the silent understanding of the body in action" (Svenaeus, 2011, p.334). In comparison to my other empirical chapters that did not use embodied interpretation, I think that this method helped locate me to others and convey this in an evocative way that made "words human" (Todres & Galvin, 2008, p.570). This method is aligned with a more aesthetic phenomenology, which seemed the most appropriate to use when describing the end of life for a child and their parents. This sought to communicate findings of a sensitive nature in a way that helps people to understand more empathically the phenomenon of dying. Overall, the adoption of these methods offered an application of qualitative findings within a 'real-world' setting that are immersed in humanising care (Yardley, 2000).

On a more practical level, this programme of work sought to be inclusive in terms of providing a way for as many people with NPC and their family carers to take part in an interview as possible. This meant that interviews were conducted through various media to the participant's preference. On average, the length of the interviews with people with NPC and/or their families was around 75 minutes. Alternative means by which the interviews were conducted did not seem to have an effect on the duration of the interview, given that some interviews over the telephone lasted 90 minutes whilst other face-to-face interviews lasted 55 minutes. On reflection, the difficulties of interviewing over the telephone manifested most prominently if the participant became upset; a common occurance given the sensitive nature of the interviews. Being present with a participant allows you to care in a way that cannot be visualised through a telephone call, such as the use of body language or the interviewer's observations of how upset the participant is and whether this impacted upon the continuation of the interview. For example, taking a break from the interview may be easier to do when in person than when speaking over the telephone. Having someone there with whom to start a new conversation, or just someone to be present whilst you compose yourself again should be considered as a possibility if preferred by the participant when conducting interviews of this nature. For future work, I think telephone interviews should be a last resort due to these reasons. Skype worked better due to being able to see each other, the reactions, the facial expressions and body language. These are important components, which play into the complexities of communicating with the personal concerns, fears, anxieties that are experienced when living with an incurable illness.

8.4.2 Implications and recommendations for clinical practice, policy and future research

8.4.2.1 Clinical implications

The findings of this programme of work have highlighted the immediate implications for improving clinical care for people living with NPC and more widely in rare disease, especially in early diagnosis and initial consultations. HCPs' training should focus on the multifaceted ways in which illness disrupts the lifeworld for people impacted by rare disease and the complexities that people face overtime. Findings suggest that more consideration should be given to concepts of time, relationships and space and how these are experienced by people living with this illness. This work suggests that more needs to be done in helping provide a sense of mobility to people impacted by NPC, helping people to feel supported when looking towards the future.

Overall, an empathic understanding of these considerations in the ways that the lifeworld is impacted would allow people to share their concerns, helping them to feel connected in the isolated world of rare disease. Frank commented "The greatest clinical gift to the ill is to appreciate them as the 'good stories' they are. In these stories there is nothing to fix, only a great deal to listen to" (Frank, 1995: p.210). This is especially pertinent in incurable conditions, where listening is one of the only ways in which we can help a person work through the situation. Todres and Galvin (2008) argued that the value of such empathic understanding, in this case through the lens of the QoL scales developed, could offer HCPs a new way of understanding knowledge, not from an "information perspective" but more in enhancing "emotional intelligence", supporting care which is more human, sensitive and ethical (Galvin et al., 2018; Todres & Galvin, 2008, p.580). The QoL scales used as a dialogue tool to assist clinicians and people impacted by NPC to talk through their story of living with illness gives a voice to their experiences, thus turning a consultation into a conversation and enacting humanised care (Galvin et al., 2018; Mannix, 2017).

As a result, the findings from this thesis suggest a re-engagement with the language and concepts used in clinical settings and within policy of people living with a LLC. As spoken about above, a re-consideration of using the concept of QoL should be carefully pondered upon in people living with incurable illnesses. This message also bleeds into other terms that hold rhetoric within society today such as: 'living well', 'dying well' and where they should be placed within the context of living with a LLC. Anxiety should not be shied away from, one should seek to understand it as to do so would resonate more with a person's lifeworld where it is common to feel isolated and cast out of a world filled with apparent 'goodness and wellness'. In anxiety comes a sense of re-generation to an authentic at-homeness, deeply embedded and connected to an interconnection of self-body-world through time and space.

Svenaeus (2000a) discussed whether phenomenological terminology or explorations of analysis such as "being-in-the world" or "at-homeness" should be used within a clinical setting, beyond the normal boundaries of phenomenological analysis. When living in the illness experience, metaphors such as these may resonate and make sense to people. This needs to be explored in more detail. However, this terminology may not be insurmountable for people to comprehend, if presented in ways that are accessible (Ahlzén, 2011; Svenaeus, 2000a), thus engaging in meaningful ways when living with incurable illness. This could be especially appropriate and relatable within palliative care, when facing the end of life; providing vocabulary that resonates in order to express their experience.

8.4.2.2 Recommendations for policy and practice:

The following recommendations can be drawn from the results of this programme of work. Although the scale and scope of these varies in ambition and challenge, they represent that which would desirable if potential barriers could be overcome.

Figure 5. Recommendations for policy and practice

- Clinicians to consider as far as they can the elements of the lifeworld to help structure consultations with individuals and families diagnosed with life-limiting conditions.
- Communicative strategies between consultants and people living with NPC, that are lifeworld-led, will routinely adopt Habermas' communicative rationality and Gavlin and colleagues' humanising care framework.
- The testimony of illness stories from patients and their parent/carer should be encouraged to help understand the experience of suffering.
- More creative measures be used in healthcare encounters to seek understanding from the experiences of children and adults living with NPC who struggle to communicate, such as writing or drawing. This should be pursued in order to understand more of the impact of the unique illness experience on their lifeworld.
- Train HCPs in how to facilitate lifeworld-led care.
- Encourage HCPs to use head-heart-hand-led care, especially with regards to death and dying, moving away from an over medicalised approach to living with a LLC.
- Support families living with NPC and rare disease more generally in feeling a sense of belonging amidst the unpredictable nature of the illness prognosis

8.4.3 Future research

Future studies should unpack the suggestions surrounding the QoL scales developed, looking at whether these would be more appropriately named as illness experience dialogue tools to aid consultations in discussing this and whether this would help people with NPC to feel more supported; a method that could be shared across rare diseases, giving much needed attention to this group. In addition to this, the development of an online community for people diagnosed with and impacted by NPC may provide an interface that is readily available for people to discuss concerns or practical advice (Newman, Lauterbach, Munson, Resnick, & Morris, 2011; Pauer et al., 2017). This may help in giving expression to and interaction between the voices of the lifeworld, providing the possibility to share experiences with people who do not need an explanation of what NPC is but can relate and understand in ways that resonate.

8.4.4 Strengths and Limitations

This programme of work has not used traditional models of illness representations to understand the illness experience but has instead been more creative and flexible in its exploration. This has been achieved through engaging with the honesty of the illness experience on a personal level and seeking understanding of new insights into times of significant challenge for the individual. This study therefore favours the illness experience account rather than being limited by traditional models of understanding the experience. Consideration needs to be given to the hermeneutic aspect of whether parents' and carers' assessment of their child's experiences of life with NPC is "accurate." I would argue that the lifeworlds of those impacted by living with NPC would merge and unify in a variety of ways during this time and thus bringing an appreciation of the carer's understanding of this would shed light on the experiences of those diagnosed, especially if the person cannot communicate due to neurological deterioration. More creative methods in terms of children or adults drawing or writing may be another way of understanding the impact of illness on the lifeworld in a new way to those with NPC.

A further consideration, which could be seen to have impacted upon the results, was the decision I made to blend the participants in chapers 4 and 5. My arguments for blending participants' stories within my analyses surrounded the issues of confidentiality and making sure that the identities of my participants were protected within the NPC UK community. The degree to which this was necessary became more unclear as I progressed in my PhD. On reflection having completed my studies, I could have approached it differently and simply asked how the participants would feel if people in the NPC UK community could identify them. However, the decision was made with good intention in respecting my participants' identities. The consequences of blending participants however, compromises the analysis to some degree, more specifically the idiographic arm of IPA. Seeing from multiple vantage points has been encouraged within IPA (Larkin et al., 2018) but the way in which this thesis reports these perspectives can be confusing in places in terms of whose voice is being presented. The lifeworlds of the person with NPC and their family carers are interweaved in parts, but still their voices are distinct. This is not always clear in chapters 4 and 5 and would be something that I would make sharper in the future when reporting a mulitperspectival IPA analysis.

With regards to the QoL tools developed, these items are not an exhaustive list and I am not claiming that what has been meaningful to people in this study is the validated 'truth' of all people with NPC. This goes against the traditional QoL scales that are statistically reliable and validated to use in practice as a clinical measure. The items are based on the specific context in which this research has been conducted and new items may unfold at a different time and with a different story. However, these questionnaires offer scope for the particular impacts on the lived experience in context of the lifeworld, providing a springboard for future research. With more time, I would have sought perspectives from HCPs involved in rare diseases work and specifically in NPC to understand their experiences of working with people who have a LLC. Understanding these interactions from a healthcare perspective may highlight more of what could be done in order to support them in understanding the existential experiences of people living with NPC, thus aiding communication between families and clinicians.

8.5 Reflections on the research process

Throughout the research process, something that I had to set aside by careful reflexive practice was my own previous assumptions. I have a level of familiarity with what it is like to care for someone living with a neurodegenerative disease and therefore have the tendency to filter narratives through my own illness experiences as a result. The disruption of disease was brought about when I was a teenager as my father was diagnosed with Alzheimer's disease at the age of 50. My father's illness lasted for over a decade, and fundamentally changed our experience of life as a family. Relationships with friends altered as people struggled to know how to be around us. The home environment became a difficult mixture of comfort and unease as debilitation took over; seeing dad have seizures threatened the sense of comfort. Conducting this research and seeing suffering in my participants, brought to the surface many aspects of my personal experiences. It not only reawakened experiences from my past that were difficult to reengage with, but it also impacted on the temporality of life as I considered how I would be if my children became ill. The portrayal of Danielle's death in chapter 6 stayed with me for a long time. Each time I bathed my daughter and plaited her hair, the felt sense of loss would come over me as I spent time thinking about her family and my family. I feel that this exposure to their narrative unveiled an understanding of their lived experience but it also caused me to become preoccupied with the essence of loss that was expressed so tangibly.

Phenomenologists have the privilege of becoming familiar with people in ways other people probably will not (Finlay, 2003). Yet, they also have the burden of taking in-depth understanding and exploration of phenomena that can be challenging to dwell upon. Unbeknownst to her, Danielle's mum challenged these emotions in me, as she told of her experiences of extended family and friends exhibiting an emotional response to the death of her daughter in a way that felt intrusive and jarring. When telling this part of her story, she explained her reaction to this, saying: "...it's not about you…this is not your story". This reaction, though straightforward in content, was a turning point for me in my research. In this drawing of the line, I was able to carry forward the mantra that "this is not my story," which helped in securing the 'felt sense' in a way that allowed me to distance myself from a sense of personal responsibility. It also helped me to see past the loss for Danielle's mum, as it did not comprise the whole story; it was intricately combined with a plethora of interrelated and interweaving responses and motivations: love, heartbreak, purpose and many others. As Frank (2000) commented:

> "Those who accept an invitation into the storytelling relation open themselves to seeing (and feeling and hearing) life differently than they normally do. Listening is not so much a willing suspension of disbelief as a willing acceptance of different beliefs and of lives in which these beliefs make sense. Whether or how a story makes sense seems not so much an analytical question as an experiential one." (p.361)

CS Lewis is quoted as saying: "We read to know we are not alone." These words increasingly permeated through me in the course of this research as I engaged with writing that resonated with my experience, providing a sense of personal acceptance. Reading the likes of Frank, Toombs and Carel has offered a place for suffering to be talked about, something I often avoided due to the uncomfortable nature of it for the recipient. These moments of connection allowed for the fact that anxiety and suffering are acceptable emotions, that they are natural and that I am not alone in feeling the deep-rooted, existential complexities that occur when one is diagnosed and lives with illness. This space for meaningful discourse should be recognised both in spite of and because of its uncomfortable nature.

8.6 Concluding remarks

This research offers support for past characteristics of the illness experience as being a "biographical disruption" (Bury, 1982), a "narrative wreckage" (Frank, 1995) whereby living with an incurable LLC irrevocably alters the lifeworld of those involved. The shared stories in this thesis have contributed in valuable ways to capturing the sense in which body-space-time impact the "all-pervasive" nature of illness (Carel, 2013). Secondly, viewing illness through the changing and shifting of these dimensions should be acknowledged as transforming one's entire being-in-the-world, thus providing a lens through which to view people's stories of living with NPC.

In this modern era, where the medicalisation of disease is pervasive and ubiquitous, finding particular expression in concepts such as 'quality of life,' 'good death' and 'living well,' the recognition of the reality of suffering and even death is avoided. Consequently, we live as a society in denial of the totality of experience, removed from engagement with a fundamental and tangible element of what it means to be human. Our inability to commune with death, to intimately know, venerate and respect it, leaves us ignorant of the meaning and authenticity that can be found in suffering and anxiety. The fullness of the lifeworld, explored in illness includes the difficult and confusing oscillation that characterises the experience; between suffering and wellbeing, hope and despair, gain and loss, strength and weakness. Growing in these understandings will provide a deeper and wider knowledge base by which care could be cultivated and delivered in a way that recognises the shared and experienced humanity in the midst of illness.

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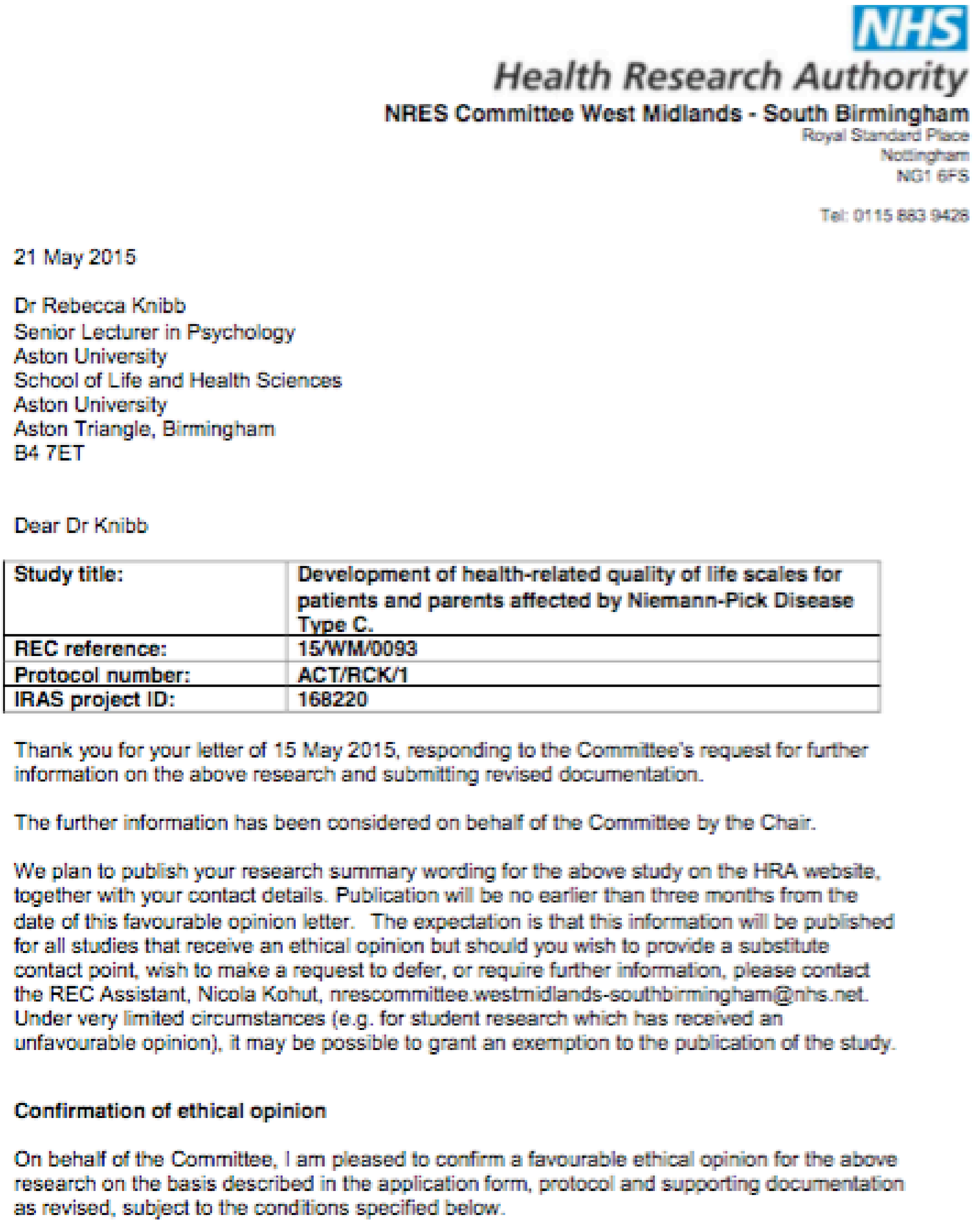
10 – Appendices

Appendix 1. NHS Ethical Approval Letter



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21 May 2015

Dr Rebecca Knibb
Senior Lecturer in Psychology
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Dear Dr Knibb

Study title:	**Development of health-related quality of life scales for patients and parents affected by Niemann-Pick Disease Type C.**
REC reference:	**15/WM/0093**
Protocol number:	**ACT/RCK/1**
IRAS project ID:	**168220**

Thank you for your letter of 15 May 2015, responding to the Committee's request for further information on the above research and submitting revised documentation.

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

We plan to publish your research summary wording for the above study on the HRA website, together with your contact details. Publication will be no earlier than three months from the date of this favourable opinion letter. The expectation is that this information will be published for all studies that receive an ethical opinion but should you wish to provide a substitute contact point, wish to make a request to defer, or require further information, please contact the REC Assistant, Nicola Kohut, nrescommittee.westmidlands-southbirmingham@nhs.net. Under very limited circumstances (e.g. for student research which has received an unfavourable opinion), it may be possible to grant an exemption to the publication of the study.

Confirmation of ethical opinion

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

Conditions of the favourable opinion

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

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

Management permission ("R&D approval") should be sought from all NHS organisations involved in the study in accordance with NHS research governance arrangements.

Guidance on applying for NHS permission for research is available in the Integrated Research Application System or at http://www.rdforum.nhs.uk.

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

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

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

Registration of Clinical Trials

All clinical trials (defined as the first four categories on the IRAS filter page) must be registered on a publically accessible database. This should be before the first participant is recruited but no later than 6 weeks after recruitment of the first participant.

There is no requirement to separately notify the REC but you should do so at the earliest opportunity e.g. when submitting an amendment. We will audit the registration details as part of the annual progress reporting process.

To ensure transparency in research, we strongly recommend that all research is registered but for non-clinical trials this is not currently mandatory.

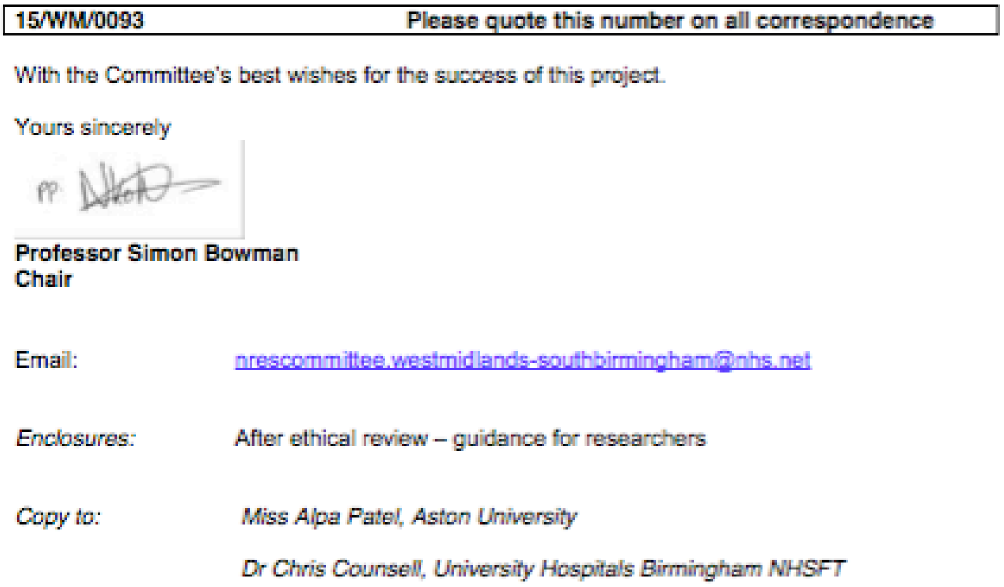
If a sponsor wishes to request a deferral for study registration within the required timeframe, they should contact hra.studyregistration@nhs.net. The expectation is that all clinical trials will be registered, however, in exceptional circumstances non registration may be permissible with prior agreement from NRES. Guidance on where to register is provided on the HRA website.

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

Ethical review of research sites

NHS sites

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



15/WM/0093 **Please quote this number on all correspondence**

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

Yours sincerely

Professor Simon Bowman
Chair

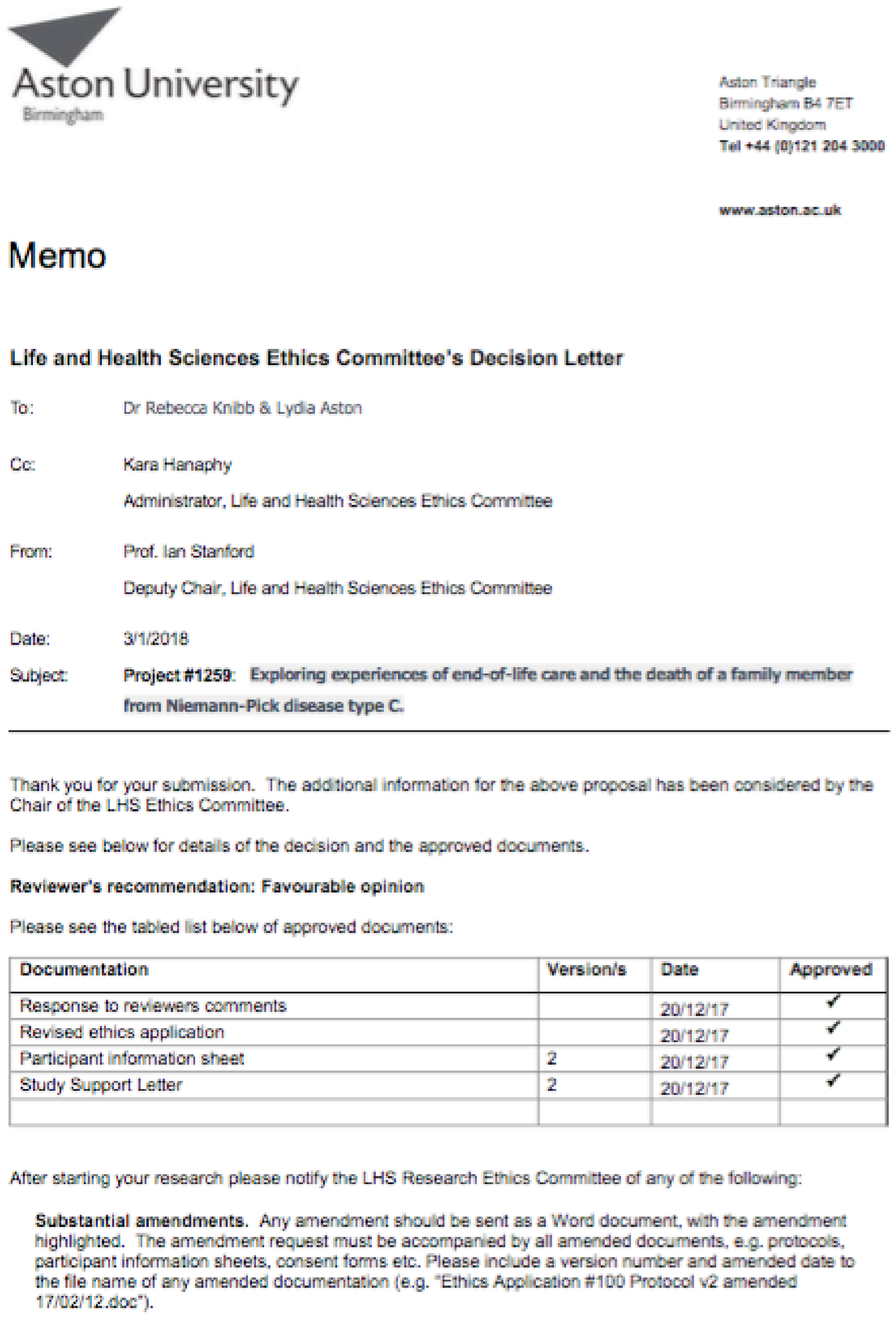
Email: nrescommittee.westmidlands-southbirmingham@nhs.net

Enclosures: After ethical review – guidance for researchers

Copy to: *Miss Alpa Patel, Aston University*

Dr Chris Counsell, University Hospitals Birmingham NHSFT

Appendix 2. Life and Health Sciences Ethics Committee's Decision Letter



Aston University
Birmingham

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Memo

Life and Health Sciences Ethics Committee's Decision Letter

To: Dr Rebecca Knibb & Lydia Aston

Cc: Kara Hanaphy
Administrator, Life and Health Sciences Ethics Committee

From: Prof. Ian Stanford
Deputy Chair, Life and Health Sciences Ethics Committee

Date: 3/1/2018

Subject: **Project #1259: Exploring experiences of end-of-life care and the death of a family member from Niemann-Pick disease type C.**

Thank you for your submission. The additional information for the above proposal has been considered by the Chair of the LHS Ethics Committee.

Please see below for details of the decision and the approved documents.

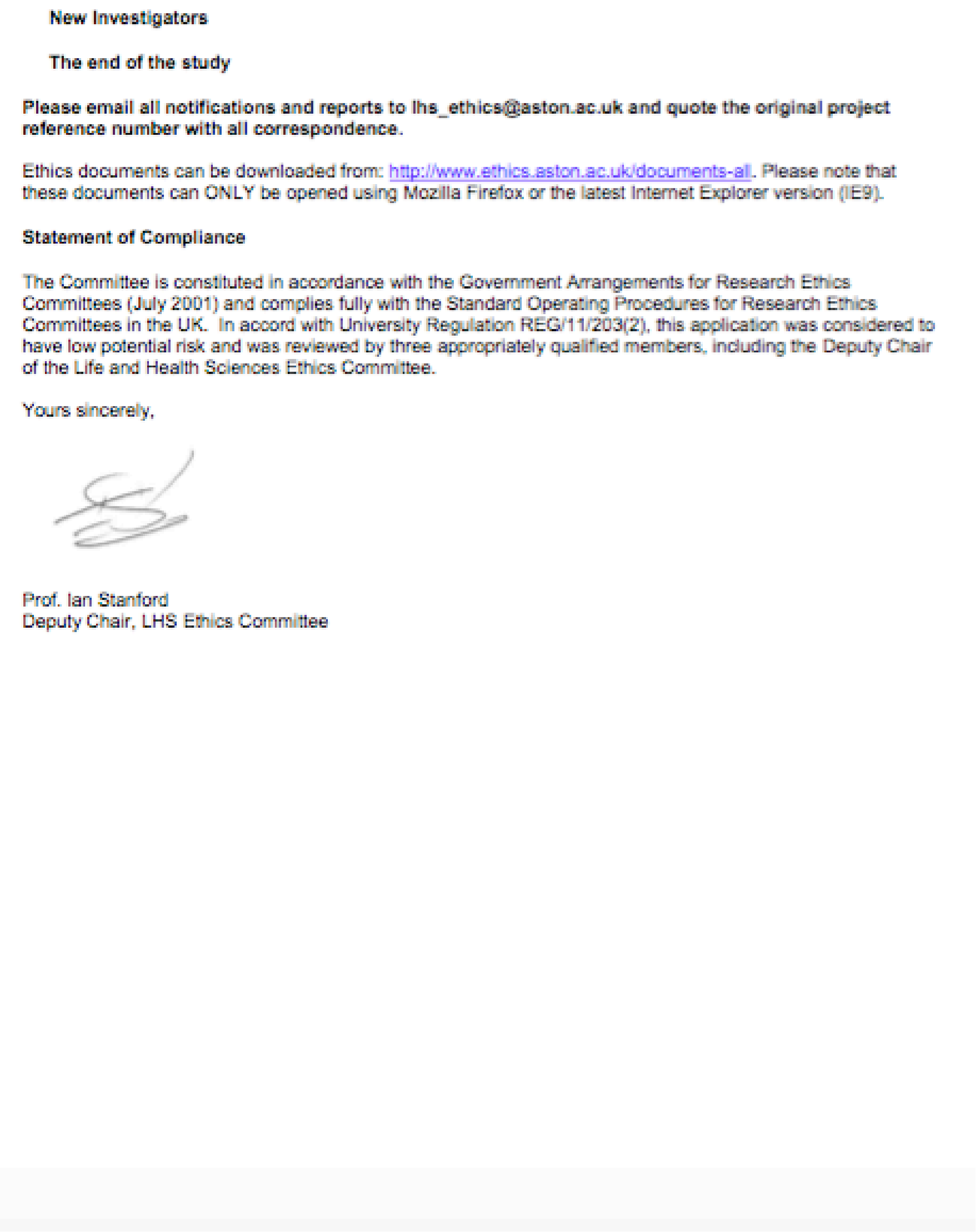
Reviewer's recommendation: Favourable opinion

Please see the tabled list below of approved documents:

Documentation	Version/s	Date	Approved
Response to reviewers comments		20/12/17	✓
Revised ethics application		20/12/17	✓
Participant information sheet	2	20/12/17	✓
Study Support Letter	2	20/12/17	✓

After starting your research please notify the LHS Research Ethics Committee of any of the following:

Substantial amendments. Any amendment should be sent as a Word document, with the amendment highlighted. The amendment request must be accompanied by all amended documents, e.g. protocols, participant information sheets, consent forms etc. Please include a version number and amended date to the file name of any amended documentation (e.g. "Ethics Application #100 Protocol v2 amended 17/02/12.doc").



New Investigators

The end of the study

Please email all notifications and reports to lhs_ethics@aston.ac.uk and quote the original project reference number with all correspondence.

Ethics documents can be downloaded from: http://www.ethics.aston.ac.uk/documents-all. Please note that these documents can ONLY be opened using Mozilla Firefox or the latest Internet Explorer version (IE9).

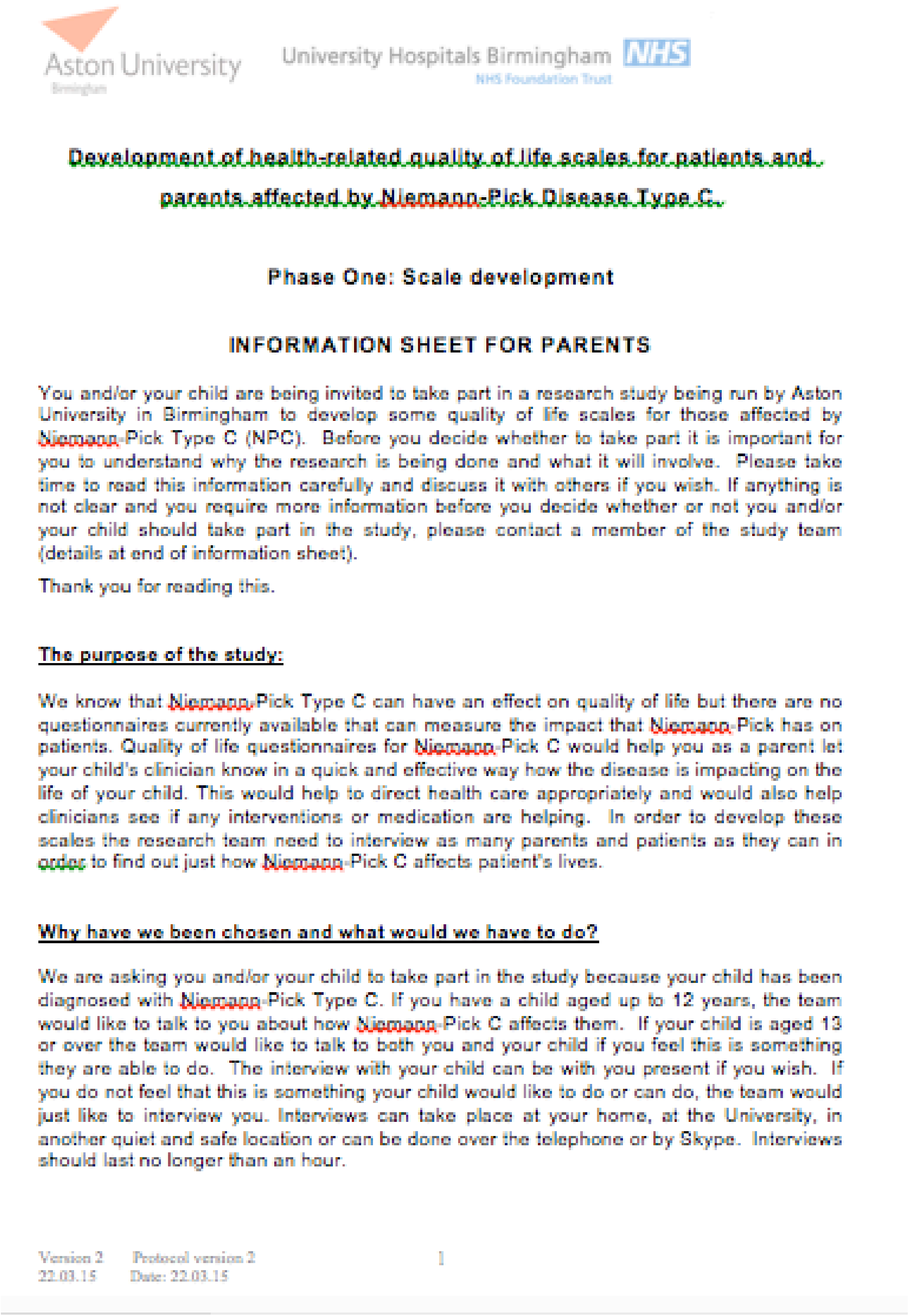
Statement of Compliance

The Committee is constituted in accordance with the Government Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK. In accord with University Regulation REG/11/203(2), this application was considered to have low potential risk and was reviewed by three appropriately qualified members, including the Deputy Chair of the Life and Health Sciences Ethics Committee.

Yours sincerely,

Prof. Ian Stanford
Deputy Chair, LHS Ethics Committee

Appendix 3. Participant information sheet for QoL studies



Aston University
Birmingham

University Hospitals Birmingham NHS
NHS Foundation Trust

Development of health-related quality of life scales for patients and parents affected by Niemann-Pick Disease Type C.

Phase One: Scale development

INFORMATION SHEET FOR PARENTS

You and/or your child are being invited to take part in a research study being run by Aston University in Birmingham to develop some quality of life scales for those affected by Niemann-Pick Type C (NPC). Before you decide whether to take part it is important for you to understand why the research is being done and what it will involve. Please take time to read this information carefully and discuss it with others if you wish. If anything is not clear and you require more information before you decide whether or not you and/or your child should take part in the study, please contact a member of the study team (details at end of information sheet).

Thank you for reading this.

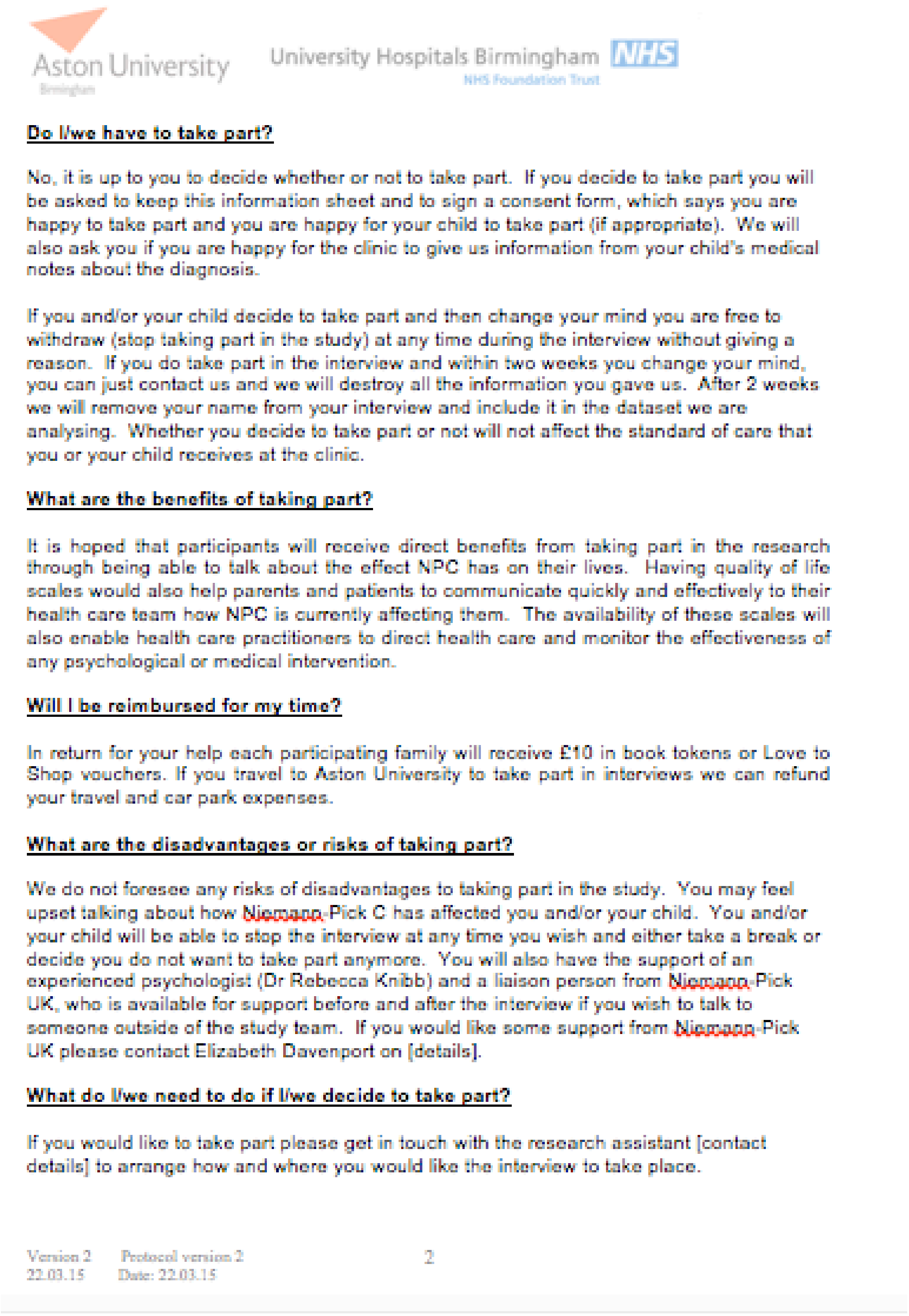
The purpose of the study:

We know that Niemann-Pick Type C can have an effect on quality of life but there are no questionnaires currently available that can measure the impact that Niemann-Pick has on patients. Quality of life questionnaires for Niemann-Pick C would help you as a parent let your child's clinician know in a quick and effective way how the disease is impacting on the life of your child. This would help to direct health care appropriately and would also help clinicians see if any interventions or medication are helping. In order to develop these scales the research team need to interview as many parents and patients as they can in order to find out just how Niemann-Pick C affects patient's lives.

Why have we been chosen and what would we have to do?

We are asking you and/or your child to take part in the study because your child has been diagnosed with Niemann-Pick Type C. If you have a child aged up to 12 years, the team would like to talk to you about how Niemann-Pick C affects them. If your child is aged 13 or over the team would like to talk to both you and your child if you feel this is something they are able to do. The interview with your child can be with you present if you wish. If you do not feel that this is something your child would like to do or can do, the team would just like to interview you. Interviews can take place at your home, at the University, in another quiet and safe location or can be done over the telephone or by Skype. Interviews should last no longer than an hour.

Version 2 22.03.15 Protocol version 2 Date: 22.03.15 1



Aston University
Birmingham

University Hospitals Birmingham NHS
NHS Foundation Trust

Do I/we have to take part?

No, it is up to you to decide whether or not to take part. If you decide to take part you will be asked to keep this information sheet and to sign a consent form, which says you are happy to take part and you are happy for your child to take part (if appropriate). We will also ask you if you are happy for the clinic to give us information from your child's medical notes about the diagnosis.

If you and/or your child decide to take part and then change your mind you are free to withdraw (stop taking part in the study) at any time during the interview without giving a reason. If you do take part in the interview and within two weeks you change your mind, you can just contact us and we will destroy all the information you gave us. After 2 weeks we will remove your name from your interview and include it in the dataset we are analysing. Whether you decide to take part or not will not affect the standard of care that you or your child receives at the clinic.

What are the benefits of taking part?

It is hoped that participants will receive direct benefits from taking part in the research through being able to talk about the effect NPC has on their lives. Having quality of life scales would also help parents and patients to communicate quickly and effectively to their health care team how NPC is currently affecting them. The availability of these scales will also enable health care practitioners to direct health care and monitor the effectiveness of any psychological or medical intervention.

Will I be reimbursed for my time?

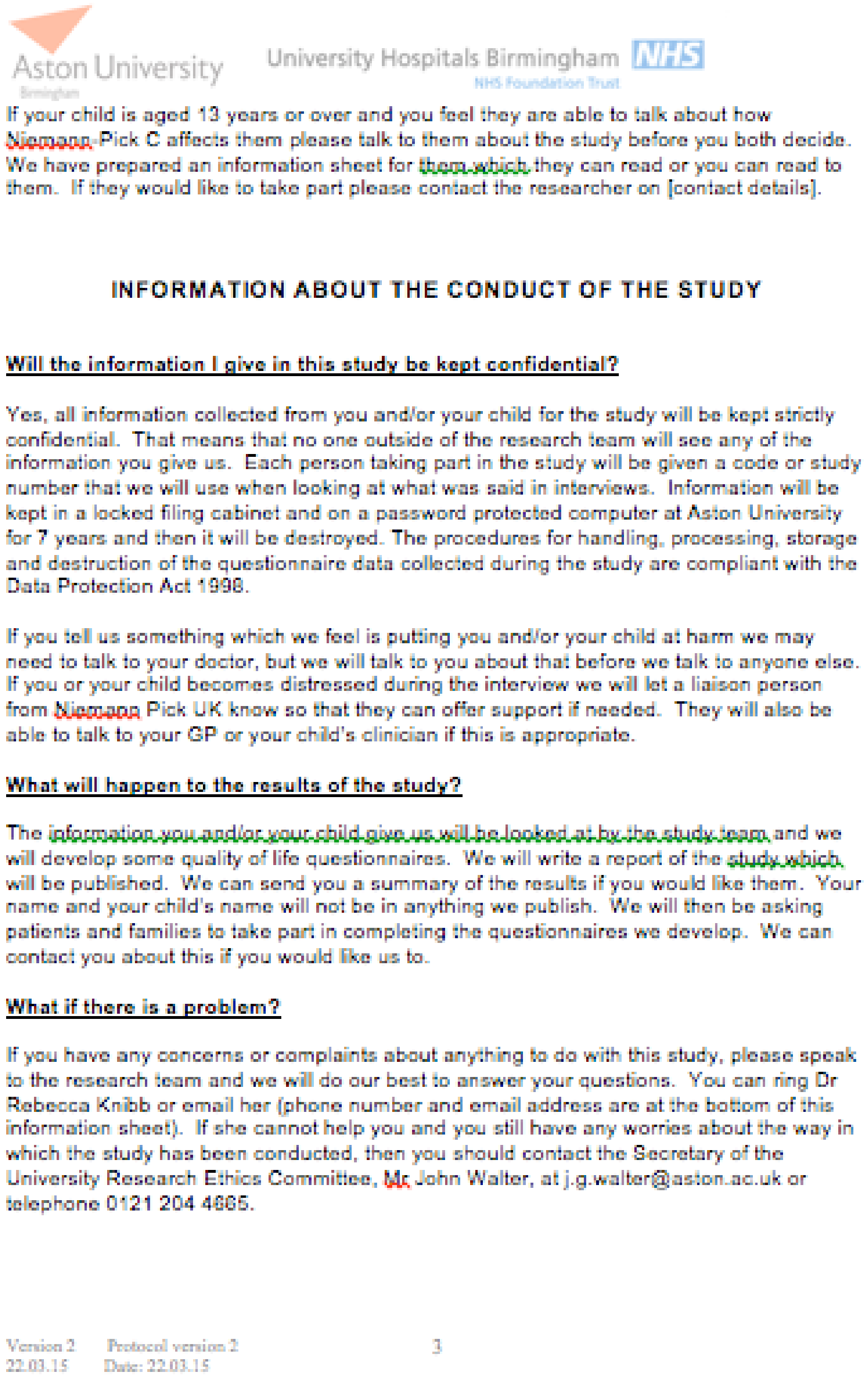
In return for your help each participating family will receive £10 in book tokens or Love to Shop vouchers. If you travel to Aston University to take part in interviews we can refund your travel and car park expenses.

What are the disadvantages or risks of taking part?

We do not foresee any risks of disadvantages to taking part in the study. You may feel upset talking about how Niemann-Pick C has affected you and/or your child. You and/or your child will be able to stop the interview at any time you wish and either take a break or decide you do not want to take part anymore. You will also have the support of an experienced psychologist (Dr Rebecca Knibb) and a liaison person from Niemann-Pick UK, who is available for support before and after the interview if you wish to talk to someone outside of the study team. If you would like some support from Niemann-Pick UK please contact Elizabeth Davenport on [details].

What do I/we need to do if I/we decide to take part?

If you would like to take part please get in touch with the research assistant [contact details] to arrange how and where you would like the interview to take place.



If your child is aged 13 years or over and you feel they are able to talk about how Niemann-Pick C affects them please talk to them about the study before you both decide. We have prepared an information sheet for them which they can read or you can read to them. If they would like to take part please contact the researcher on [contact details].

INFORMATION ABOUT THE CONDUCT OF THE STUDY

Will the information I give in this study be kept confidential?

Yes, all information collected from you and/or your child for the study will be kept strictly confidential. That means that no one outside of the research team will see any of the information you give us. Each person taking part in the study will be given a code or study number that we will use when looking at what was said in interviews. Information will be kept in a locked filing cabinet and on a password protected computer at Aston University for 7 years and then it will be destroyed. The procedures for handling, processing, storage and destruction of the questionnaire data collected during the study are compliant with the Data Protection Act 1998.

If you tell us something which we feel is putting you and/or your child at harm we may need to talk to your doctor, but we will talk to you about that before we talk to anyone else. If you or your child becomes distressed during the interview we will let a liaison person from Niemann Pick UK know so that they can offer support if needed. They will also be able to talk to your GP or your child's clinician if this is appropriate.

What will happen to the results of the study?

The information you and/or your child give us will be looked at by the study team and we will develop some quality of life questionnaires. We will write a report of the study which will be published. We can send you a summary of the results if you would like them. Your name and your child's name will not be in anything we publish. We will then be asking patients and families to take part in completing the questionnaires we develop. We can contact you about this if you would like us to.

What if there is a problem?

If you have any concerns or complaints about anything to do with this study, please speak to the research team and we will do our best to answer your questions. You can ring Dr Rebecca Knibb or email her (phone number and email address are at the bottom of this information sheet). If she cannot help you and you still have any worries about the way in which the study has been conducted, then you should contact the Secretary of the University Research Ethics Committee, Mr John Walter, at j.g.walter@aston.ac.uk or telephone 0121 204 4665.

Who has reviewed the study?

This study has been looked by the West Midlands and South Birmingham Ethics Committee for NHS research and given a favourable opinion. These are a group of people who check research to make sure that it protects the safety, rights, wellbeing and dignity of anyone who takes part.

Can I get more information?

If you are interested in taking part in this study but would like some more information before you decide, please talk to Dr Rebecca Knibb or email us. Or please feel free to talk to your child's consultant at your clinic. If you would like to talk to an independent person about taking part in this study or about research in general in Psychology at Aston University please contact the Director of the Aston Research Centre for Children's and Young People's Health, Professor Helen Pattison, on h.pattison@aston.ac.uk.

If you would like independent advice on any aspect of this study, you can also contact the PALS (Patient Advice and Liaison Service) at the University Hospitals Birmingham NHS Trust on 0121 371 3280.

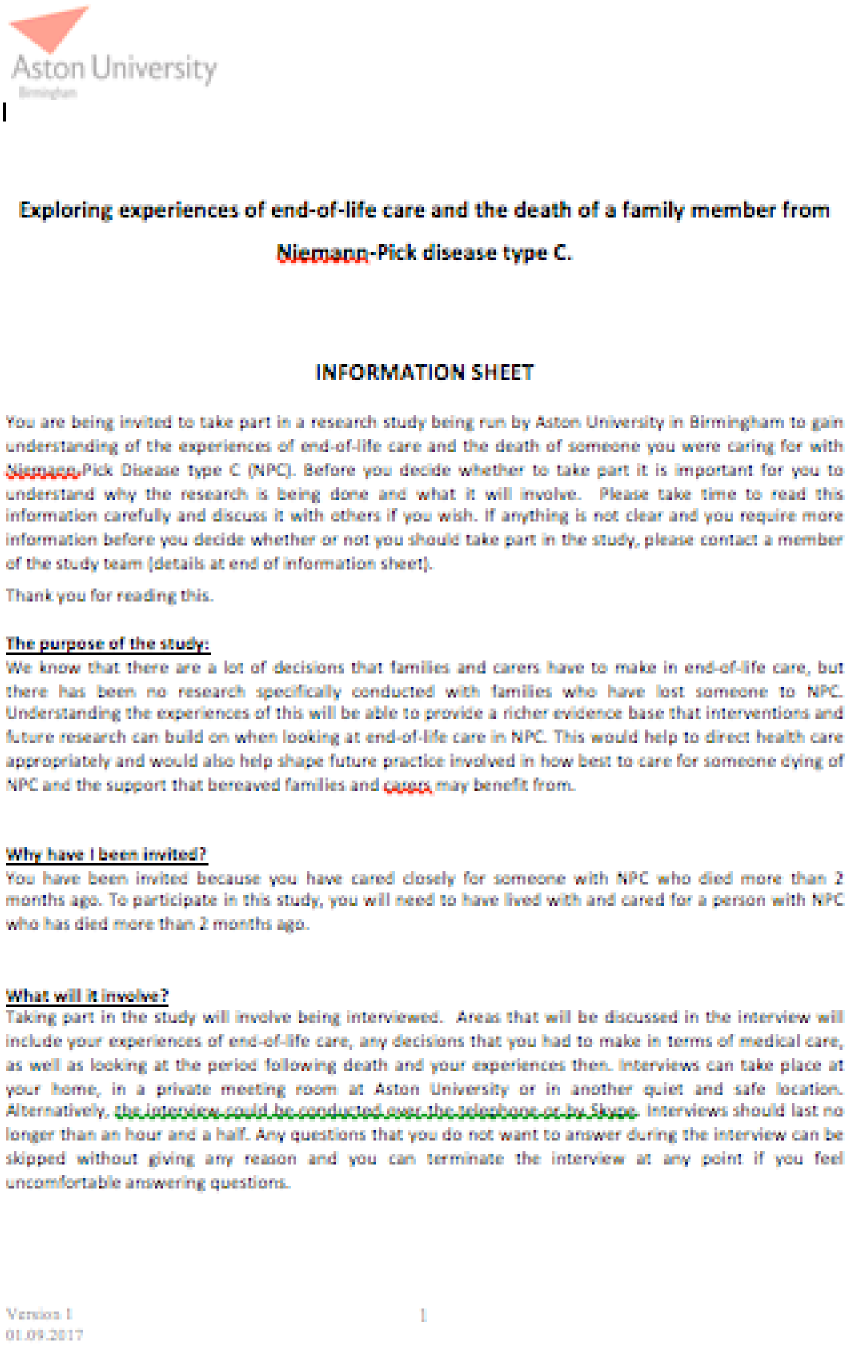
Thank you for taking time to read this information sheet.

Yours sincerely

Dr Rebecca Knibb (Chief Investigator)
Health Psychologist
School of Life and Health Sciences
Aston University
Birmingham
B4 7ET
Email: r.knibb@aston.ac.uk
Telephone: 0121 204 3402

Lydia Aston (co-investigator)
School of Life and Health Sciences
Aston University
Birmingham
B4 7ET
Email: astonlr@aston.ac.uk

Appendix 4. Participant information sheet for end-of-life care and dying experience study



Aston University
Birmingham

Exploring experiences of end-of-life care and the death of a family member from Niemann-Pick disease type C.

INFORMATION SHEET

You are being invited to take part in a research study being run by Aston University in Birmingham to gain understanding of the experiences of end-of-life care and the death of someone you were caring for with Niemann-Pick Disease type C (NPC). Before you decide whether to take part it is important for you to understand why the research is being done and what it will involve. Please take time to read this information carefully and discuss it with others if you wish. If anything is not clear and you require more information before you decide whether or not you should take part in the study, please contact a member of the study team (details at end of information sheet).

Thank you for reading this.

The purpose of the study:

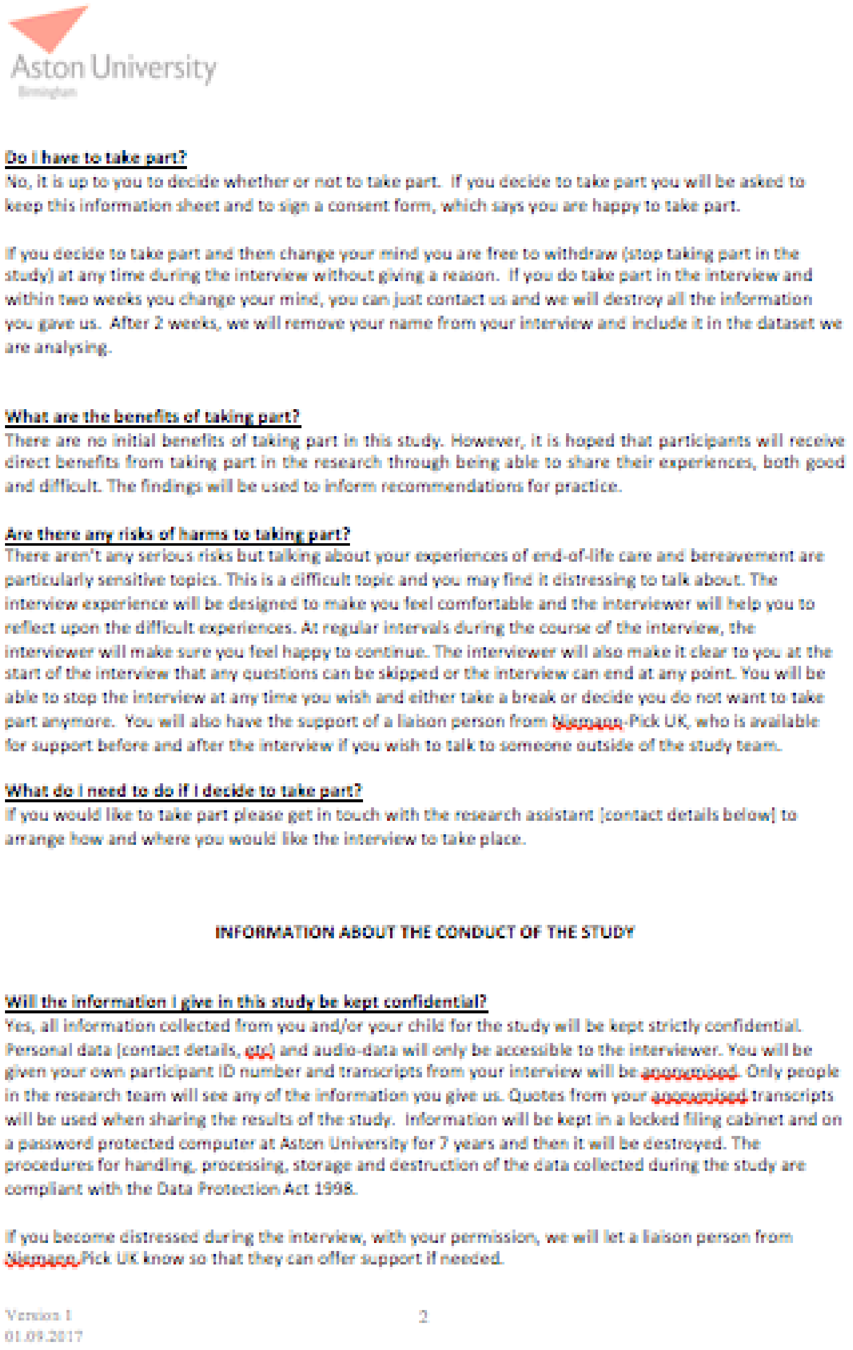
We know that there are a lot of decisions that families and carers have to make in end-of-life care, but there has been no research specifically conducted with families who have lost someone to NPC. Understanding the experiences of this will be able to provide a richer evidence base that interventions and future research can build on when looking at end-of-life care in NPC. This would help to direct health care appropriately and would also help shape future practice involved in how best to care for someone dying of NPC and the support that bereaved families and carers may benefit from.

Why have I been invited?

You have been invited because you have cared closely for someone with NPC who died more than 2 months ago. To participate in this study, you will need to have lived with and cared for a person with NPC who has died more than 2 months ago.

What will it involve?

Taking part in the study will involve being interviewed. Areas that will be discussed in the interview will include your experiences of end-of-life care, any decisions that you had to make in terms of medical care, as well as looking at the period following death and your experiences then. Interviews can take place at your home, in a private meeting room at Aston University or in another quiet and safe location. Alternatively, the interview could be conducted over the telephone or by Skype. Interviews should last no longer than an hour and a half. Any questions that you do not want to answer during the interview can be skipped without giving any reason and you can terminate the interview at any point if you feel uncomfortable answering questions.



Do I have to take part?

No, it is up to you to decide whether or not to take part. If you decide to take part you will be asked to keep this information sheet and to sign a consent form, which says you are happy to take part.

If you decide to take part and then change your mind you are free to withdraw (stop taking part in the study) at any time during the interview without giving a reason. If you do take part in the interview and within two weeks you change your mind, you can just contact us and we will destroy all the information you gave us. After 2 weeks, we will remove your name from your interview and include it in the dataset we are analysing.

What are the benefits of taking part?

There are no initial benefits of taking part in this study. However, it is hoped that participants will receive direct benefits from taking part in the research through being able to share their experiences, both good and difficult. The findings will be used to inform recommendations for practice.

Are there any risks of harms to taking part?

There aren't any serious risks but talking about your experiences of end-of-life care and bereavement are particularly sensitive topics. This is a difficult topic and you may find it distressing to talk about. The interview experience will be designed to make you feel comfortable and the interviewer will help you to reflect upon the difficult experiences. At regular intervals during the course of the interview, the interviewer will make sure you feel happy to continue. The interviewer will also make it clear to you at the start of the interview that any questions can be skipped or the interview can end at any point. You will be able to stop the interview at any time you wish and either take a break or decide you do not want to take part anymore. You will also have the support of a liaison person from Niemann-Pick UK, who is available for support before and after the interview if you wish to talk to someone outside of the study team.

What do I need to do if I decide to take part?

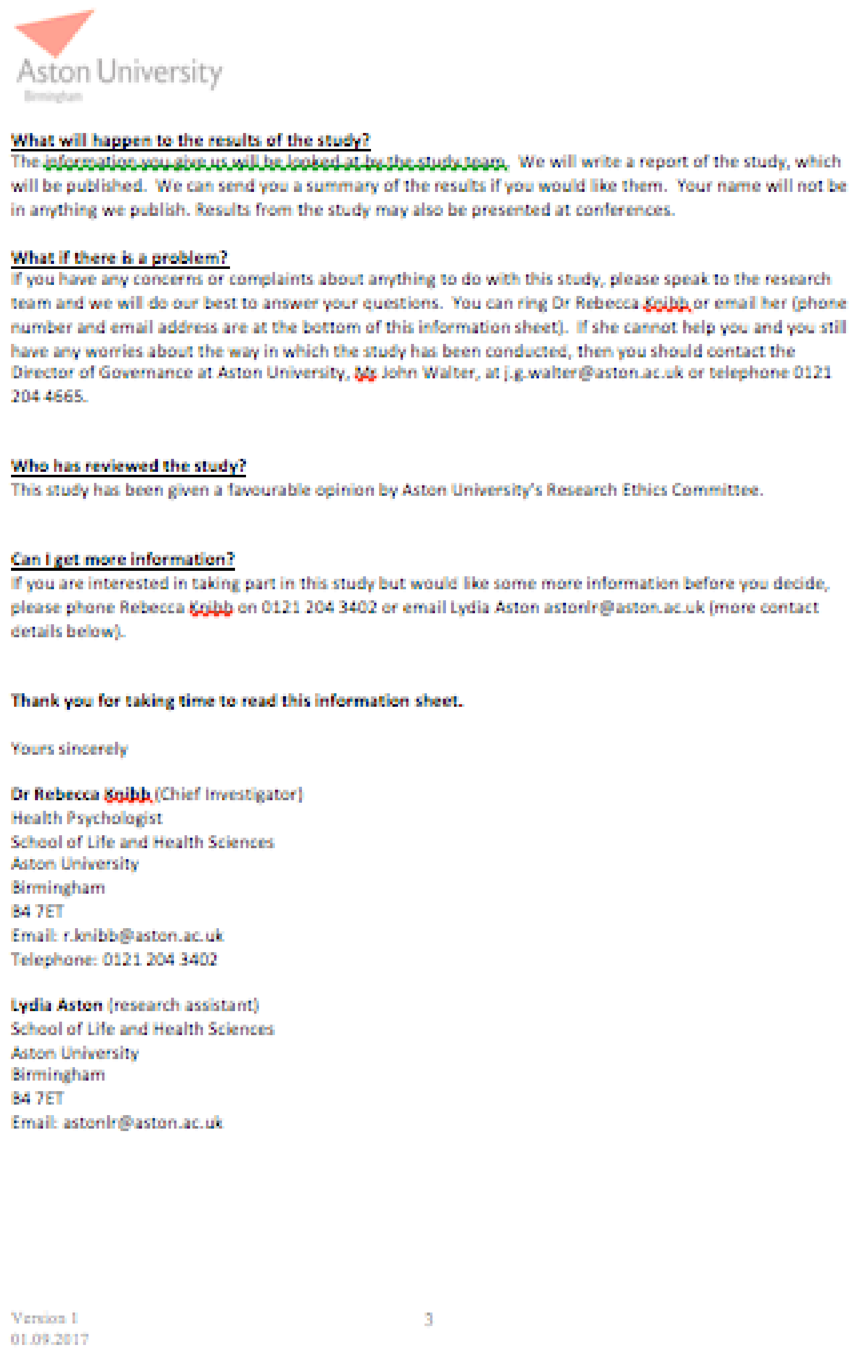
If you would like to take part please get in touch with the research assistant [contact details below] to arrange how and where you would like the interview to take place.

INFORMATION ABOUT THE CONDUCT OF THE STUDY

Will the information I give in this study be kept confidential?

Yes, all information collected from you and/or your child for the study will be kept strictly confidential. Personal data (contact details, etc) and audio-data will only be accessible to the interviewer. You will be given your own participant ID number and transcripts from your interview will be anonymised. Only people in the research team will see any of the information you give us. Quotes from your anonymised transcripts will be used when sharing the results of the study. Information will be kept in a locked filing cabinet and on a password protected computer at Aston University for 7 years and then it will be destroyed. The procedures for handling, processing, storage and destruction of the data collected during the study are compliant with the Data Protection Act 1998.

If you become distressed during the interview, with your permission, we will let a liaison person from Niemann-Pick UK know so that they can offer support if needed.



Aston University
Birmingham

What will happen to the results of the study?

The information you give us will be looked at by the study team. We will write a report of the study, which will be published. We can send you a summary of the results if you would like them. Your name will not be in anything we publish. Results from the study may also be presented at conferences.

What if there is a problem?

If you have any concerns or complaints about anything to do with this study, please speak to the research team and we will do our best to answer your questions. You can ring Dr Rebecca Knibb or email her (phone number and email address are at the bottom of this information sheet). If she cannot help you and you still have any worries about the way in which the study has been conducted, then you should contact the Director of Governance at Aston University, Mr John Walter, at j.g.walter@aston.ac.uk or telephone 0121 204 4665.

Who has reviewed the study?

This study has been given a favourable opinion by Aston University's Research Ethics Committee.

Can I get more information?

If you are interested in taking part in this study but would like some more information before you decide, please phone Rebecca Knibb on 0121 204 3402 or email Lydia Aston astonlr@aston.ac.uk (more contact details below).

Thank you for taking time to read this information sheet.

Yours sincerely

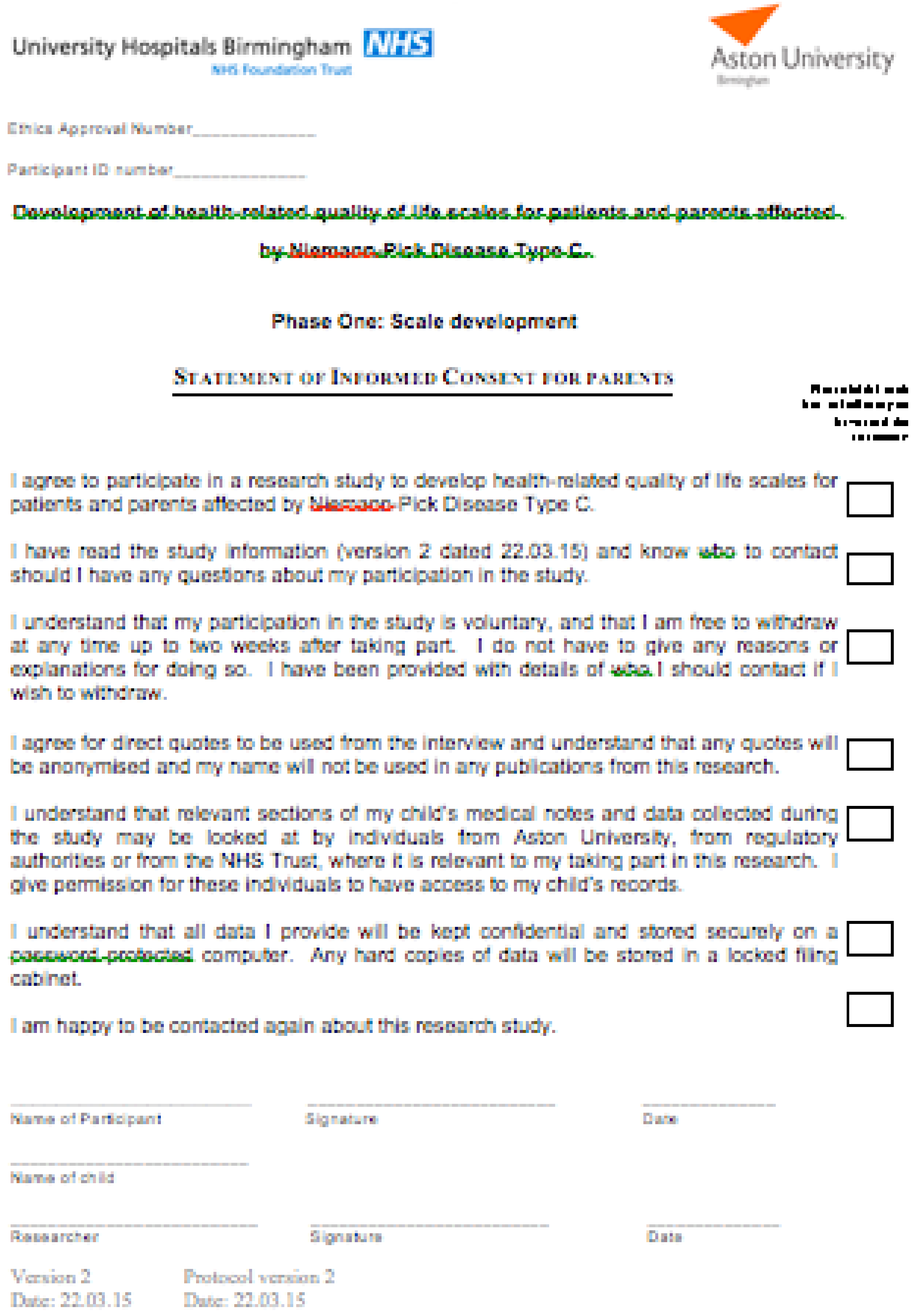
Dr Rebecca Knibb (Chief Investigator)
Health Psychologist
School of Life and Health Sciences
Aston University
Birmingham
B4 7ET
Email: r.knibb@aston.ac.uk
Telephone: 0121 204 3402

Lydia Aston (research assistant)
School of Life and Health Sciences
Aston University
Birmingham
B4 7ET
Email: astonlr@aston.ac.uk

Version 1
01.09.2017

3

Appendix 5. Consent sheet for QoL study



University Hospitals Birmingham NHS
NHS Foundation Trust

Aston University
Birmingham

Ethics Approval Number_______________

Participant ID number_______________

Development of health-related quality of life scales for patients and parents affected by Niemann-Pick Disease Type C.

Phase One: Scale development

STATEMENT OF INFORMED CONSENT FOR PARENTS

[illegible]

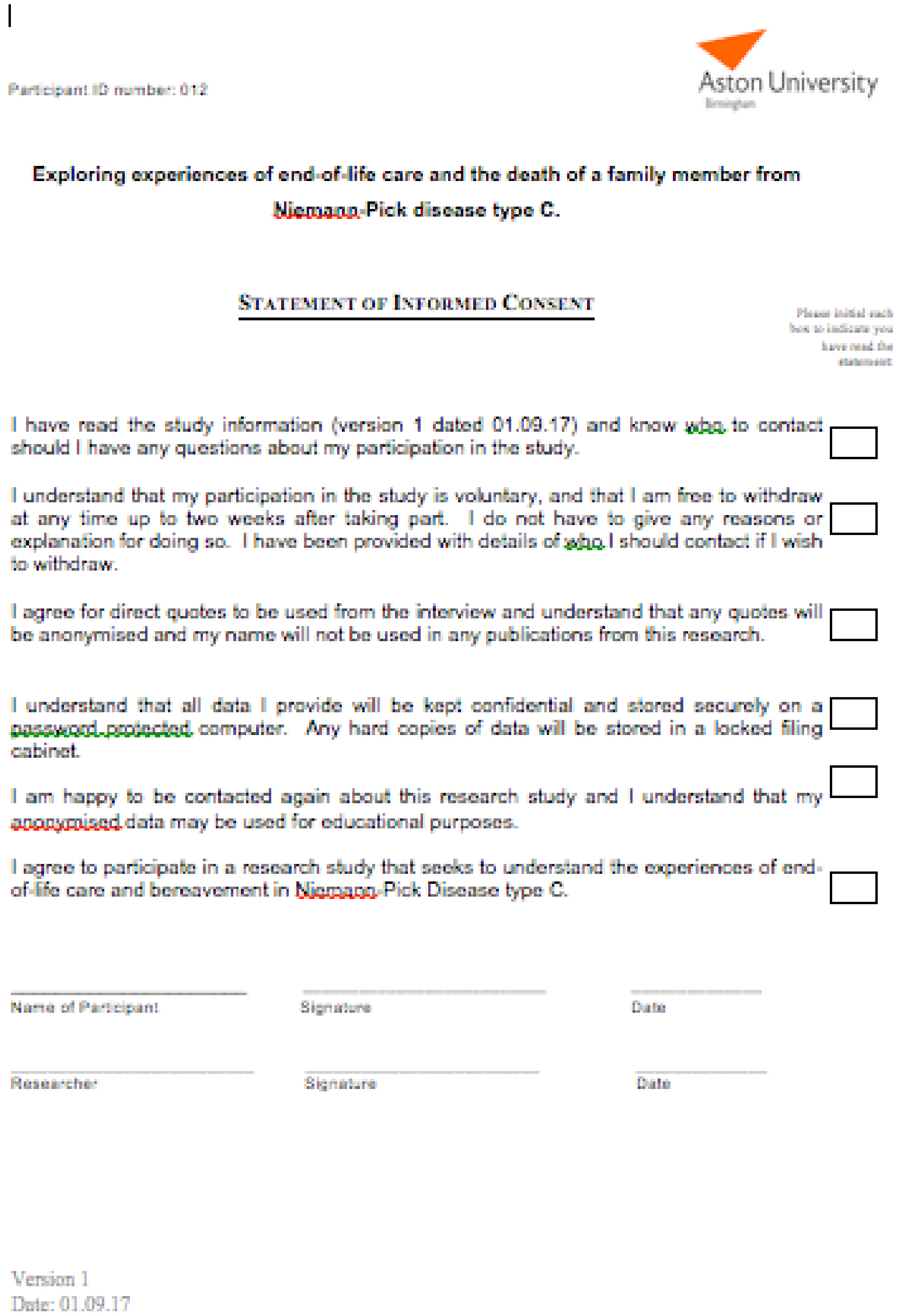
I agree to participate in a research study to develop health-related quality of life scales for patients and parents affected by Niemann-Pick Disease Type C.	☐
I have read the study information (version 2 dated 22.03.15) and know who to contact should I have any questions about my participation in the study.	☐
I understand that my participation in the study is voluntary, and that I am free to withdraw at any time up to two weeks after taking part. I do not have to give any reasons or explanations for doing so. I have been provided with details of who I should contact if I wish to withdraw.	☐
I agree for direct quotes to be used from the interview and understand that any quotes will be anonymised and my name will not be used in any publications from this research.	☐
I understand that relevant sections of my child's medical notes and data collected during the study may be looked at by individuals from Aston University, from regulatory authorities or from the NHS Trust, where it is relevant to my taking part in this research. I give permission for these individuals to have access to my child's records.	☐
I understand that all data I provide will be kept confidential and stored securely on a password protected computer. Any hard copies of data will be stored in a locked filing cabinet.	☐
I am happy to be contacted again about this research study.	☐

____________________	____________________	____________
Name of Participant	Signature	Date
____________________		
Name of child		
____________________	____________________	____________
Researcher	Signature	Date

Version 2
Date: 22.03.15

Protocol version 2
Date: 22.03.15

Appendix 6. Consent form for end-of-life and dying experience study



Participant ID number: 012

Aston University

Exploring experiences of end-of-life care and the death of a family member from Niemann-Pick disease type C.

STATEMENT OF INFORMED CONSENT

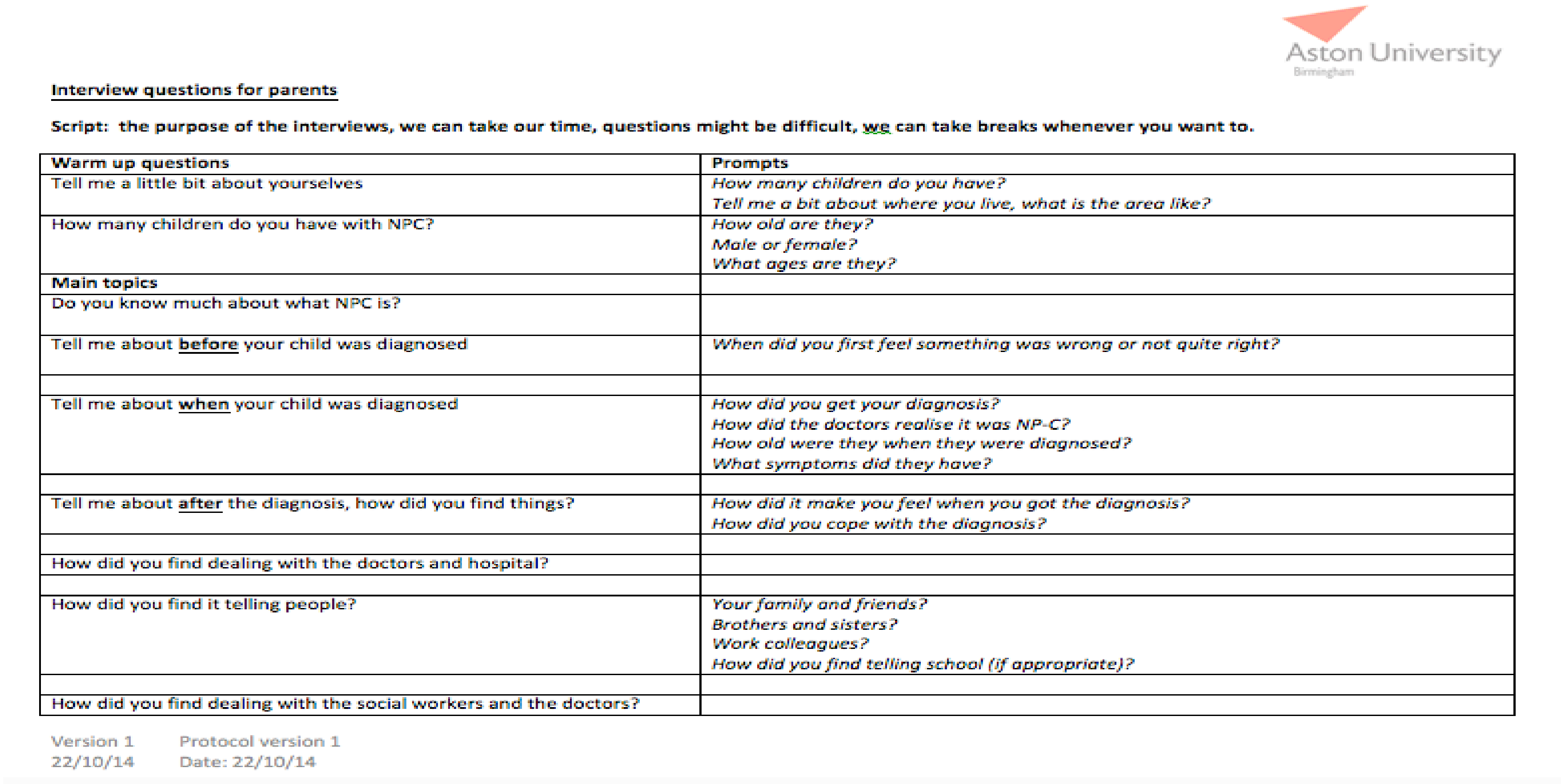
Please initial each box to indicate you have read the statement

I have read the study information (version 1 dated 01.09.17) and know who to contact should I have any questions about my participation in the study.	☐
I understand that my participation in the study is voluntary, and that I am free to withdraw at any time up to two weeks after taking part. I do not have to give any reasons or explanation for doing so. I have been provided with details of who I should contact if I wish to withdraw.	☐
I agree for direct quotes to be used from the interview and understand that any quotes will be anonymised and my name will not be used in any publications from this research.	☐
I understand that all data I provide will be kept confidential and stored securely on a password protected computer. Any hard copies of data will be stored in a locked filing cabinet.	☐
I am happy to be contacted again about this research study and I understand that my anonymised data may be used for educational purposes.	☐
I agree to participate in a research study that seeks to understand the experiences of end-of-life care and bereavement in Niemann-Pick Disease type C.	☐

Name of Participant	Signature	Date
Researcher	Signature	Date

Version 1
Date: 01.09.17

Appendix 7. Interview schdule for parents of children with NPC



Aston University
Birmingham

Interview questions for parents

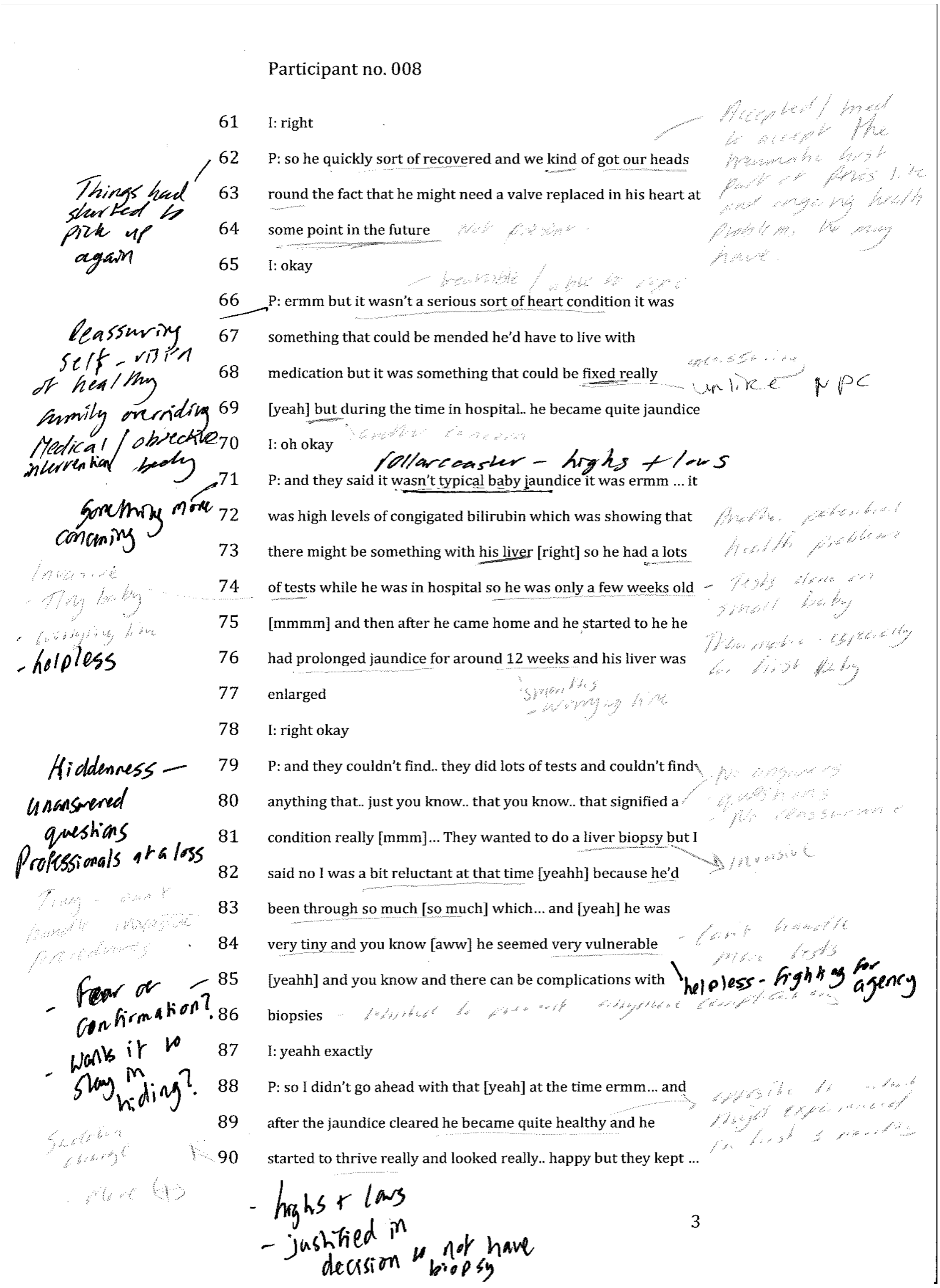
Script: the purpose of the interviews, we can take our time, questions might be difficult, we can take breaks whenever you want to.

Warm up questions	**Prompts**
Tell me a little bit about yourselves	*How many children do you have?* *Tell me a bit about where you live, what is the area like?*
How many children do you have with NPC?	*How old are they?* *Male or female?* *What ages are they?*
Main topics	
Do you know much about what NPC is?	
Tell me about **before** your child was diagnosed	*When did you first feel something was wrong or not quite right?*
Tell me about **when** your child was diagnosed	*How did you get your diagnosis?* *How did the doctors realise it was NP-C?* *How old were they when they were diagnosed?* *What symptoms did they have?*
Tell me about **after** the diagnosis, how did you find things?	*How did it make you feel when you got the diagnosis?* *How did you cope with the diagnosis?*
How did you find dealing with the doctors and hospital?	
How did you find it telling people?	*Your family and friends?* *Brothers and sisters?* *Work colleagues?* *How did you find telling school (if appropriate)?*
How did you find dealing with the social workers and the doctors?	

Version 1 Protocol version 1
22/10/14 Date: 22/10/14

Tell me about how things are ***now***?	*What symptoms does your child have now?* *What can they do for themselves?* *What do you have to do to help them?* *Is there anything they miss out on or have had to give up?* *Is there anything you have had to miss out on or give up?*
Tell me how things are on a day-to-day basis?	*What is a typical day for you?* *What is a typical night for you? How well do you sleep/does your child sleep?*
How does your child cope with their symptoms?	*Is there anything they do that helps them with their symptoms?* *Is there anything you do to help them cope?* *What sort of things do you do to help you or your child?*
How do they find taking their medication?	*What medication are they on?* *Does the medication help them?* *Do you do anything you think makes taking the medication easier for your child?*
Does your child take any complimentary medication?	*Does this help at all?*
Does your child have physio- or hydro-therapy?	*Does this help at all?*
Have you had to make any changes to where you live?	*Have you had to have any special equipment?* *Have you had to change your living environment/house/access/car?*
Have there been any impact on finances?	*Have you had to give up work?*
Do you have any outside help with looking after your child?	*How do you find this?/Would you like this if it were available?*
Is there anything you need that you are not getting?	

Appendix 8. Example of annotated transcript



Participant no. 008

I: right

P: so he quickly sort of recovered and we kind of got our heads round the fact that he might need a valve replaced in his heart at some point in the future

I: okay

P: ermm but it wasn't a serious sort of heart condition it was something that could be mended he'd have to live with medication but it was something that could be fixed really [yeah] but during the time in hospital.. he became quite jaundice

I: oh okay

P: and they said it wasn't typical baby jaundice it was ermm … it was high levels of congigated bilirubin which was showing that there might be something with his liver [right] so he had a lots of tests while he was in hospital so he was only a few weeks old [mmmm] and then after he came home and he started to he he had prolonged jaundice for around 12 weeks and his liver was enlarged

I: right okay

P: and they couldn't find.. they did lots of tests and couldn't find anything that.. just you know.. that you know.. that signified a condition really [mmm]… They wanted to do a liver biopsy but I said no I was a bit reluctant at that time [yeahh] because he'd been through so much [so much] which… and [yeah] he was very tiny and you know [aww] he seemed very vulnerable [yeahh] and you know and there can be complications with biopsies

I: yeahh exactly

P: so I didn't go ahead with that [yeah] at the time ermm… and after the jaundice cleared he became quite healthy and he started to thrive really and looked really.. happy but they kept …

3

Things had started to pick up again

Reassuring self - vision of healthy family overriding Medical / objective intervention body

Something more concerning

Invasive
- Tiny baby
- worrying him
- helpless

Hiddenness — unanswered questions
Professionals at a loss

Tiny - can't handle invasive procedures

- Fear of confirmation?
- Wants it to stay in hiding?

Section change

More (+)

Not present -

- treatable / able to repair

Another concern

rollarcoaster - highs + lows

Something - worrying him

- justified to prevent unnecessary complications

- highs + lows
- justified in decision to not have biopsy

Accepted / tried to accept the traumatic first part of this life and ongoing health problem, he may have.

[illegible] unlike NPC

Another potential health problem

- Tests done on small baby

Traumatic - especially for first baby

- No answers
- questions
- No reassurance

Invasive

- Can't handle more tests

helpless - fighting for agency

[illegible] to [illegible] major experienced in first 3 months

Participant no. 008

he was under the care of Dr McClean at St. James' hospital but he kept saying theres something that nots quite right because he shouldn't have an enlarged liver… other health professionals and health visitors were saying he looks really well and he looks really healthy [yeah yeah].. other children with liver problems look poorly.. they have tubes and and itching and skin problems and he didn't have any of that ermm his liver just remained large but he was kind of developing quite well [yeah yeah] looking very health [yeah] erm so I kinda put it to the back of my mind erm and it was his heart problem that we kinda focused on really that time ermm.. and you just kept going.. having to go every 6 months to the liver clinic and when he was 3 nearly 4 years old they said perhaps it might be time to do a biopsy to … to try and get the bottom of it really [okay] because they wondered whether it was his heart.. regurgitations of his heart going back to his liver.. that might be damaging the liver so we kinda thought we should really check it out erm… and then we went along to … what we thought was a normal clinic and… there were lots of children there and we were kept waiting for a long time … and erm we were eventually the last patients to be seen walked into a room full of professionals and they just gave us the devastating news [ohhhh] just to had to just pick him up and take him home with me

I: yeah

P: and… after after .. he was just short of his 4th birthday actually at that point [awww]… so we were introducted to Jackie who was the niemann pick nurse [yeah yeah] and… and she came out to see us and … kinda explained what was happening really and how it had occurred through a faulty gene which we never heard of or associated conditions you know in that way before

Still lots of questions
No explanations

- Repetition
- child hungry [illegible] ever
- Not completely [illegible] yet even though he appeared healthy

- People who you trust

- Comparisons to images of babies who are poorly in hospital

Mixed messages

Looked normal

- Confusion or contradictions [illegible]
- No answers

- Moving forwards?
- Fear of diagnosis?

Couldn't turn away easily?

Long time before finding out / allowed [illegible] to happen - Really be [illegible] Time to Find answers to these questions

Tried to focus on the future

focus more on (+) of what HCPs have said.

an possible explanation

It was time

No previous knowledge

Frustrating

Kept to be seen

Special case

helpless

Biggest puzzle waiting to be solved

- Frustrated
- HCPs could not answer questions

get him away from it all
- pick up body

- Fascination
- objective diseased 'body'

No prior information
- Didn't know anything about the disease

HCPs had not explained

- Lost / Didn't know
- Didn't know implications

- HCP's communication

HCPs kept her in the dark

Appendix 9. Interview schedule for patients with NPC

Aston University

Interview questions for patients

Script: the purpose of the interviews, we can take our time, questions might be difficult, we can take breaks whenever you want to.

Warm up questions	
How are you today?	
Have you had a good day?	
What have you been doing today?	
Main topics	
Do you know much about what NPC is?	
Can you tell me about **your NPC**?	
How long have you had it for? Can you remember?	
Can you remember a time when you didn't have it?	
How does it affect you?	*Does your NPC stop you doing anything?* *Is there anything you struggle with because of your NPC?* *Do you have good days and bad days?*
Tell me about your day today? How has NPC affected you today?	*What has been good about today?* *What has been not so good about today?* *Is there anything that has troubled you today?* *Is there anything that you have struggled with today?* *How have you coped with that? What did you do to make things better?* *Is there anything your mum/dad/partner/friend etc do to help?*
Generally is there anything you struggle with because of your NP-C?	*What do you regularly struggle with?* *What about today or yesterday?*

Version 1 Protocol version 1
22/10/14 Date: 22/10/14

Is there anything you do to make you feel better?	
Is there anything that makes you feel worse?	
What bothers you most about your NPC?	
How do you sleep at night?	
How do you find taking your medication?	
How do you find eating?	
How did you find dealing with the doctors and hospital/hospice?	
What school/college/work do you go to? (if applicable)	
What is it like going to school/college/work? (if applicable)	
Do you have any special friends at school/college/work? (if applicable)	*Do you talk to them about your NPC?*
How do you feel about your NP-C?	
Is there anything you have had to miss out on or give up because of your NPC?	
Is there anywhere you go to have time for yourself?	
Is there anything that would make life better for you?	
Is there anything else that you would like to say that we haven't already talked about?	

What school does your child go to? (if applicable)	
How does your child find school? (if applicable)	
Do they have any special friends at school? (if applicable)	
How do you think your child feels about their NP-C (if applicable)	*Have you noticed anything different in their behaviour?*
How does it make **you feel**?	*How do you cope with it?*
Has NPC changed your relationships with your family or friends?	
Has NPC changed your relationships with your community and around where you live?	
Is there anything that would make life better for your child and for you?	
Are you making any plans for the future?	
Is there anything else that you would like to say that we haven't already talked about?	

Appendix 10. Interview schedule for end-of-life and dying experience study

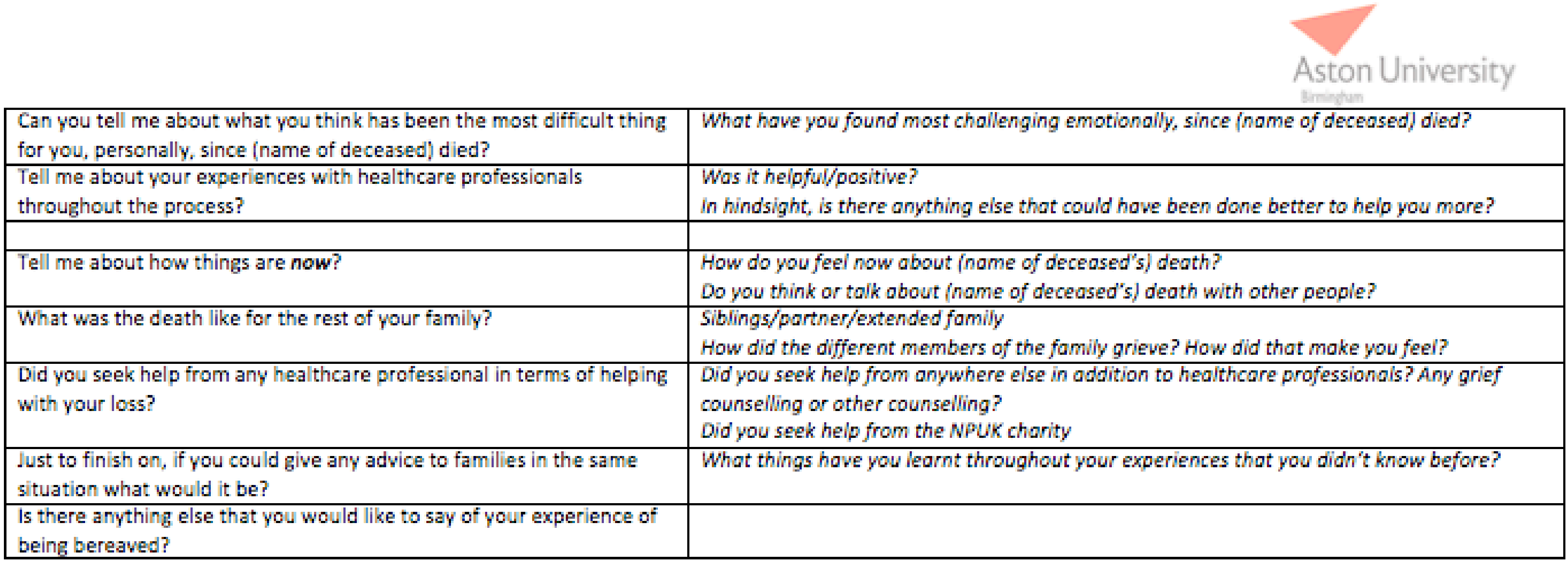
Interview questions for people who have been bereaved

Note to committee: This schedule will be used as a guide and not an explicit script as it is uncertain what direction the participants will take the interview. Before the interview commences, participants will be reminded of the purpose of the interviews, and that the questions might be difficult, but that we can take our time and have breaks at any point during the interview.

Warm up questions	**Prompts**
Tell me a little bit about yourselves	*Tell me about your family* *Relationship to (name of deceased)?* *Do you have any other children?*
When was (name of deceased) diagnosed with NPC?	*How old were they?* *Length of illness?* *Did you know much about NPC before diagnosis?*
About deceased and dying experience	
Can you tell me about when (name of deceased) passed away?	*How long ago did this happen?* *How old were they when they died?*
Can you tell me about your experiences leading up to the death of (name of deceased)?	*Were they admitted to hospital in the week leading up to their death?* *How would you describe the level of care (name of deceased) received before their death?*
Can you tell me about any decisions you had to make in terms of medical care when (name of deceased) was dying?	*Can you tell me about the interactions you had with healthcare professionals?* *Do you feel that you were able to make the decisions you felt were best for (name of deceased)?* *Was there an end of life care plan in place?* *Where did (name of deceased) pass away?*
Can you tell me about how you found the dying experience?	*How did the dying experience feel to you?*

Version 1
01/09/17

	Were you present when they passed away? *Do you feel you were able to say good-bye?*
Period following death	
Can you tell me what things were like for you, personally, when it first happened?	*How long did you feel that way?* *What was going on in your mind/body?* *Did those feelings change as time progressed?*
What was it like at home/work after losing (name of deceased)?	*Did home feel different?* *How did evenings and weekends feel?* *Did places/spaces feel different in meaning? Become more significant?*
Can you tell me about how it was when you told your friends and family about your loss?	*How was it being around people after it had happened?* *How did family/friends react to (name of deceased) death?* *Did it change over time?* *If so, in what ways did it change?* *What would you say was most difficult for you as a family during this early period?*
What, if anything, did you find consoling during this time?	*What helped you through your grief?*
Can you tell me more about your grieving experiences?	*Did you feel like it was a process? Was it something that you were aware of?* *Were there things happening or that you did that you didn't really notice at the time?*
How do you feel you looked as a family from the outside?	*Would other people know what was happening to them?* *How did other people react to your grieve?*
Can you tell me the ways in which this experience has changed you?	*Has it changed how you view the world?* *Do you think you see things differently now?* *Has it affected how you view the present and the future?*
Has this loss changed the way you live now?	*Do you see things from a different perspective and if so, can you tell me more about that?*



Can you tell me about what you think has been the most difficult thing for you, personally, since (name of deceased) died?	*What have you found most challenging emotionally, since (name of deceased) died?*
Tell me about your experiences with healthcare professionals throughout the process?	*Was it helpful/positive?* *In hindsight, is there anything else that could have been done better to help you more?*
Tell me about how things are ***now***?	*How do you feel now about (name of deceased's) death?* *Do you think or talk about (name of deceased's) death with other people?*
What was the death like for the rest of your family?	*Siblings/partner/extended family* *How did the different members of the family grieve? How did that make you feel?*
Did you seek help from any healthcare professional in terms of helping with your loss?	*Did you seek help from anywhere else in addition to healthcare professionals? Any grief counselling or other counselling?* *Did you seek help from the NPUK charity*
Just to finish on, if you could give any advice to families in the same situation what would it be?	*What things have you learnt throughout your experiences that you didn't know before?*
Is there anything else that you would like to say of your experience of being bereaved?	